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A network of disorders and disease genes linked by known disorder–gene associations offers a platform to explore in a single graph-theoretic framework all known phenotype and disease gene associations, indicating the common genetic origin of many diseases. Genes associated with similar disorders show both higher likelihood of physical interactions between their products and higher expression profiling similarity for their transcripts, supporting the existence of distinct disease-specific functional modules. We find that essential human genes are likely to encode hub proteins and are expressed widely in most tissues. This suggests that disease genes also would play a central role in the human interactome. In contrast, we find that the vast majority of disease genes are nonessential and show no tendency to encode hub proteins, and their expression pattern indicates that they are localized in the functional periphery of the network. A selection-based model explains the observed difference between essential and disease genes and also suggests that diseases caused by somatic mutations should not be peripheral, a prediction we confirm for cancer genes.

biological networks | complex networks | human genetics | systems biology | diseasome

Decades-long efforts to map human disease loci, at first genetically and later physically (1), followed by recent positional cloning of many disease genes (2) and genome-wide association studies (3), have generated an impressive list of disorder–gene association pairs (4, 5). In addition, recent efforts to map the protein–protein interactions in humans (6, 7), together with efforts to curate an extensive map of human metabolism (8) and regulatory networks offer increasingly detailed maps of the relationships between different disease genes. Most of the successful studies building on these new approaches have focused, however, on a single disease, using network-based tools to gain a better understanding of the relationship between the genes implicated in a selected disorder (9).

Here we take a conceptually different approach, exploring whether human genetic disorders and the corresponding disease genes might be related to each other at a higher level of cellular and organismal organization. Support for the validity of this approach is provided by examples of genetic disorders that arise from mutations in more than a single gene (locus heterogeneity). For example, Zellweger syndrome is caused by mutations in any of at least 11 genes, all associated with peroxisome biogenesis (10). Similarly, there are many examples of different mutations in the same gene (allelic heterogeneity) giving rise to phenotypes currently classified as different disorders. For example, mutations in *TP53* have been linked to 11 clinically distinguishable cancer-related disorders (11). Given the highly interlinked internal organization of the cell (12–17), it should be possible to improve the single gene–single disorder approach by developing a conceptual framework to link systematically all genetic disorders (the human "disease phenome") with the complete list of disease genes (the "disease genome"), resulting in a global view of the "diseasome," the combined set of all known disorder/disease gene associations.

Results

Construction of the Diseasome. We constructed a bipartite graph consisting of two disjoint sets of nodes. One set corresponds to all known genetic disorders, whereas the other set corresponds to all known disease genes in the human genome (Fig. 1). A disorder and a gene are then connected by a link if mutations in that gene are implicated in that disorder. The list of disorders, disease genes, and associations between them was obtained from the Online Mendelian Inheritance in Man (OMIM; ref. 18), a compendium of human disease genes and phenotypes. As of December 2005, this list contained 1,284 disorders and 1,777 disease genes. OMIM initially focused on monogenic disorders but in recent years has expanded to include complex traits and the associated genetic mutations that confer susceptibility to these common disorders (18). Although this history introduces some biases, and the disease gene record is far from complete, OMIM represents the most complete and up-to-date repository of all known disease genes and the disorders they confer. We manually classified each disorder into one of 22 disorder classes based on the physiological system affected [see supporting information (SI) *Text*, SI Fig. 5, and SI Table 1 for details].

Starting from the diseasome bipartite graph we generated two biologically relevant network projections (Fig. 1). In the "human disease network" (HDN) nodes represent disorders, and two disorders are connected to each other if they share at least one gene in which mutations are associated with both disorders (Figs. 1 and 2*a*). In the "disease gene network" (DGN) nodes represent disease genes, and two genes are connected if they are associated with the same disorder (Figs. 1 and 2*b*). Next, we discuss the potential of these networks to help us understand and represent in a single framework all known disease gene and phenotype associations.

Properties of the HDN. If each human disorder tends to have a distinct and unique genetic origin, then the HDN would be disconnected into many single nodes corresponding to specific disorders or grouped into small clusters of a few closely related disorders. In contrast, the obtained HDN displays many connections between both individual disorders and disorder classes (Fig. 2*a*). Of 1,284 disorders, 867 have at least one link to other disorders, and 516 disorders form a giant component, suggesting that the genetic origins of most diseases, to some extent, are shared with other diseases. The number of genes associated with a disorder, s, has a broad distribution (see SI Fig. 6*a*), indicating that most disorders relate to a few disease genes, whereas a handful of phenotypes, such as deafness ($s = 41$), leukemia ($s = 37$), and colon cancer ($s = 34$), relate to dozens of genes (Fig. 2*a*). The degree (k) distribution of HDN (SI Fig. 6*b*) indicates that most disorders are linked to only

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Abbreviations: DGN, disease gene network; HDN, human disease network; GO, Gene Ontology; OMIM, Online Mendelian Inheritance in Man; PCC, Pearson correlation coefficient.

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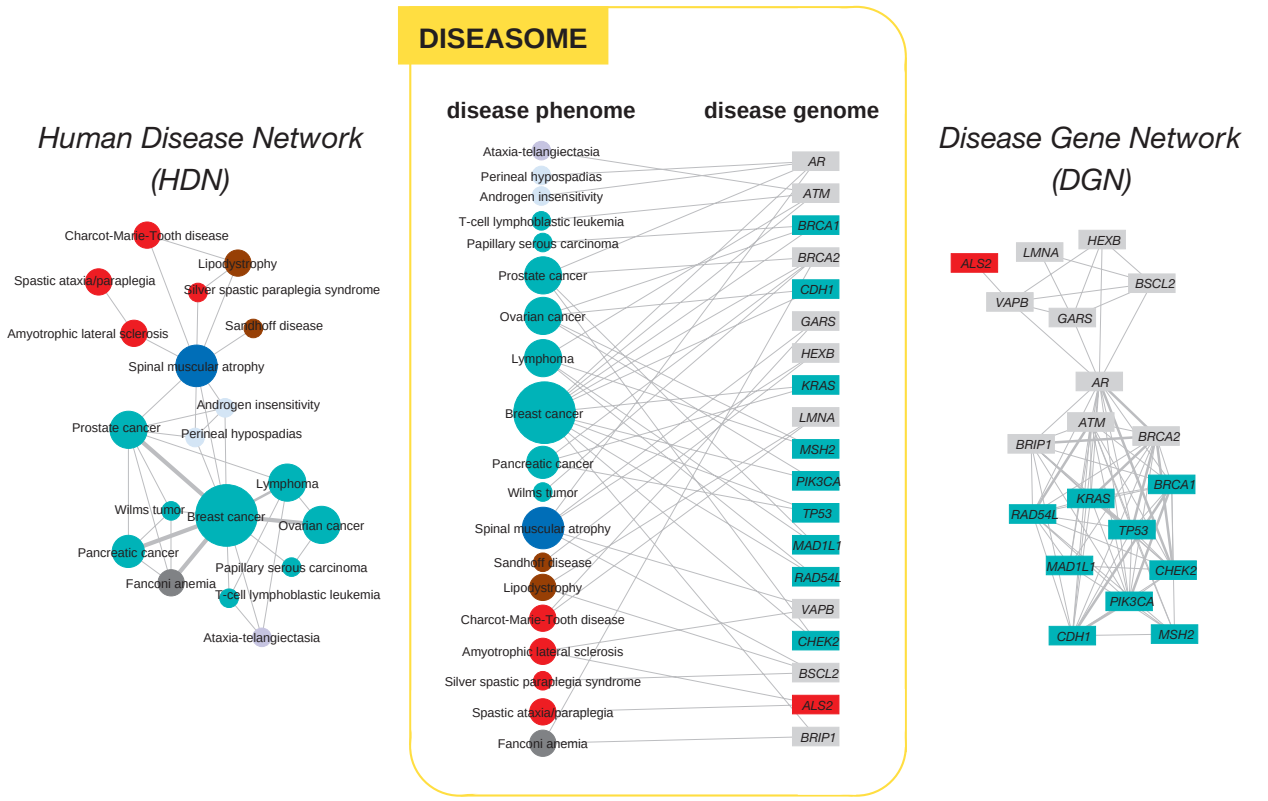

Fig. 1. Construction of the diseasome bipartite network. (*Center*) A small subset of OMIM-based disorder–disease gene associations (18), where circles and rectangles correspond to disorders and disease genes, respectively. A link is placed between a disorder and a disease gene if mutations in that gene lead to the specific disorder. The size of a circle is proportional to the number of genes participating in the corresponding disorder, and the color corresponds to the disorder class to which the disease belongs. (*Left*) The HDN projection of the diseasome bipartite graph, in which two disorders are connected if there is a gene that is implicated in both. The width of a link is proportional to the number of genes that are implicated in both diseases. For example, three genes are implicated in both breast cancer and prostate cancer, resulting in a link of weight three between them. (*Right*) The DGN projection where two genes are connected if they are involved in the same disorder. The width of a link is proportional to the number of diseases with which the two genes are commonly associated. A full diseasome bipartite map is provided as SI Fig. 13.

a few other disorders, whereas a few phenotypes such as colon cancer (linked to $k = 50$ other disorders) or breast cancer ($k = 30$) represent hubs that are connected to a large number of distinct disorders. The prominence of cancer among the most connected disorders arises in part from the many clinically distinct cancer subtypes tightly connected with each other through common tumor repressor genes such as *TP53* and *PTEN*.

Although the HDN layout was generated independently of any knowledge on disorder classes, the resulting network is naturally and visibly clustered according to major disorder classes. Yet, there are visible differences between different classes of disorders. Whereas the large cancer cluster is tightly interconnected due to the many genes associated with multiple types of cancer (*TP53*, *KRAS*, *ERBB2*, *NF1*, etc.) and includes several diseases with strong predisposition to cancer, such as Fanconi anemia and ataxia telangiectasia, metabolic disorders do not appear to form a single distinct cluster but are underrepresented in the giant component and overrepresented in the small connected components (Fig. 2*a*). To quantify this difference, we measured the locus heterogeneity of each disorder class and the fraction of disorders that are connected to each other in the HDN (see *SI Text*). We find that cancer and neurological disorders show high locus heterogeneity and also represent the most connected disease classes, in contrast with metabolic, skeletal, and multiple disorders that have low genetic heterogeneity and are the least connected (SI Fig. 7).

Properties of the DGN. In the DGN, two disease genes are connected if they are associated with the same disorder, providing a complementary, gene-centered view of the diseasome. Given that the links signify related phenotypic association between two genes, they represent a measure of their phenotypic relatedness, which could be used in future studies, in conjunction with protein–protein interactions (6, 7, 19), transcription factor-promoter interactions (20), and metabolic reactions (8), to discover novel genetic interactions. In the DGN, 1,377 of 1,777 disease genes are connected to other disease genes, and 903 genes belong to a giant component (Fig. 2*b*). Whereas the number of genes involved in multiple diseases decreases rapidly (SI Fig. 6*d*; light gray nodes in Fig. 2*b*), several disease genes (e.g., *TP53*, *PAX6*) are involved in as many as 10 disorders, representing major hubs in the network.

Functional Clustering of HDN and DGN. To probe how the topology of the HDN and GDN deviates from random, we randomly shuffled the associations between disorders and genes, while keeping the number of links per each disorder and disease gene in the bipartite network unchanged. Interestingly, the average size of the giant component of 10^4 randomized disease networks is 643 ± 16, significantly larger than 516 ($P < 10^{-4}$; for details of statistical analyses of the results reported hereafter, see *SI Text*), the actual size of the HDN (SI Fig. 6*c*). Similarly, the average size of the giant component from randomized gene networks is $1{,}087 \pm 20$ genes, significantly larger than 903 ($P < 10^{-4}$), the actual size of the DGN (SI Fig. 6*e*). These differences suggest important pathophysiological clustering of disorders and disease genes. Indeed, in the actual networks disorders (genes) are more likely linked to disorders (genes) of the same disorder class. For example, in the HDN there

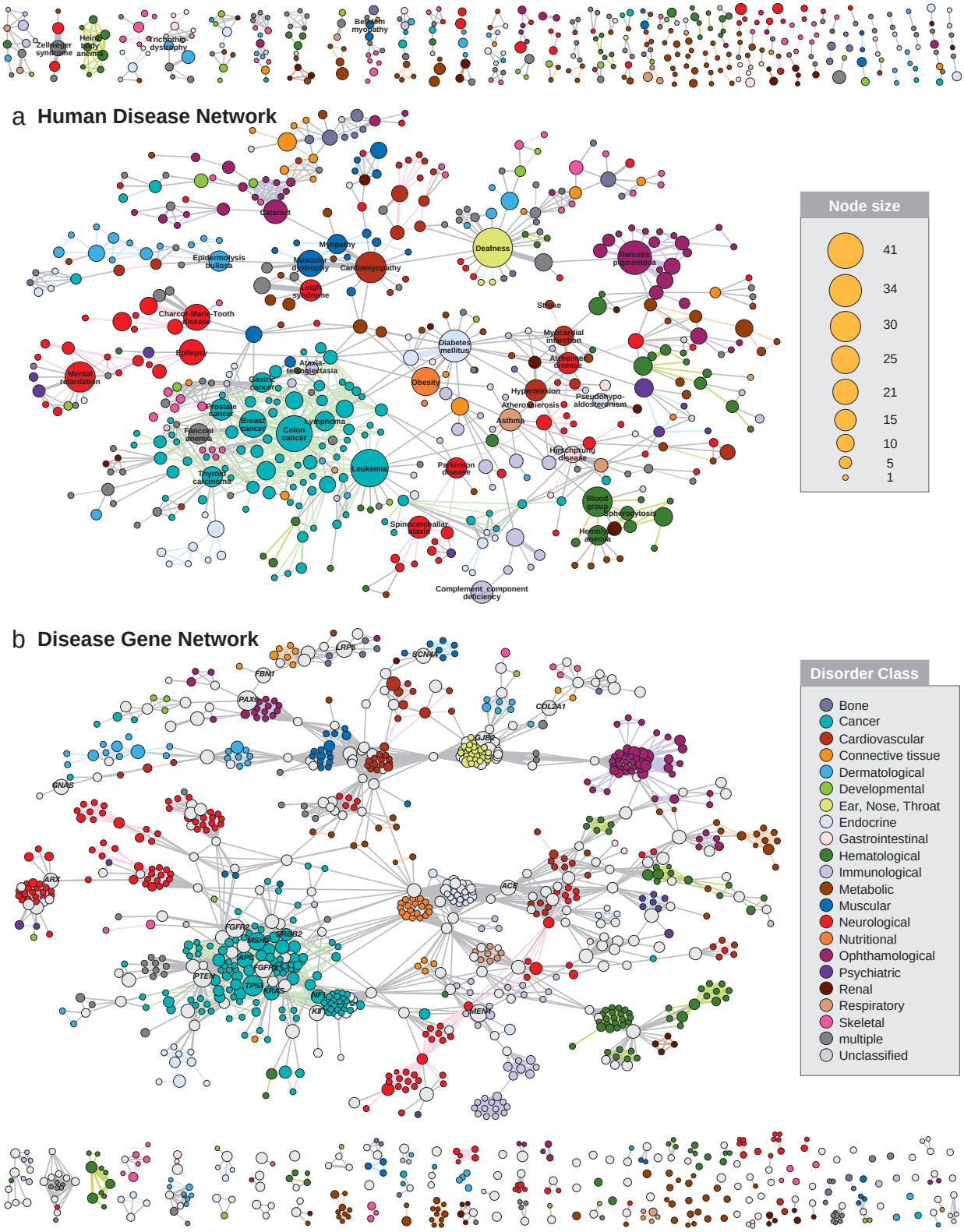

Fig. 2. The HDN and the DGN. (*a*) In the HDN, each node corresponds to a distinct disorder, colored based on the disorder class to which it belongs, the name of the 22 disorder classes being shown on the right. A link between disorders in the same disorder class is colored with the corresponding dimmer color and links connecting different disorder classes are gray. The size of each node is proportional to the number of genes participating in the corresponding disorder (see key), and the link thickness is proportional to the number of genes shared by the disorders it connects. We indicate the name of disorders with >10 associated genes, as well as those mentioned in the text. For a complete set of names, see SI Fig. 13. (*b*) In the DGN, each node is a gene, with two genes being connected if they are implicated in the same disorder. The size of each node is proportional to the number of disorders in which the gene is implicated (see key). Nodes are light gray if the corresponding genes are associated with more than one disorder class. Genes associated with more than five disorders, and those mentioned in the text, are indicated with the gene symbol. Only nodes with at least one link are shown.

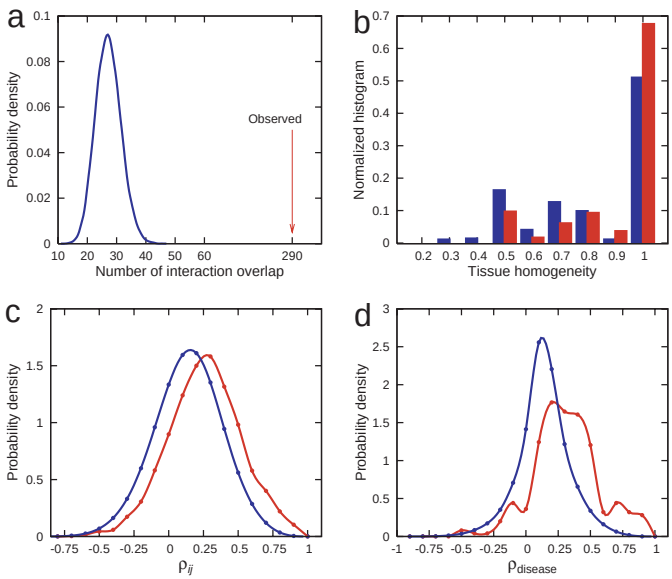

Fig. 3. Characterizing the disease modules. (*a*) Number of observed physical interactions between the products of genes within the same disorder (red arrow) and the distribution of the expected number of interactions for the random control (blue) ($P < 10^{-6}$). (*b*) Distribution of the tissue-homogeneity of a disorder (red). Random control (blue) with the same number of genes chosen randomly is shown for comparison. (*c*) The distribution of PCC ρ_{ij} values of the expression profiles of each disease gene pair that belongs to the same disorder (red) and the control (blue), representing the PCC distribution between all gene pairs ($P < 10^{-6}$). (*d*) Distribution of the average PCC between expression profiles of all genes associated with the same disorder (red) is also shifted toward higher values than the random control (blue) with the same number of genes chosen randomly ($P < 10^{-6}$).

are 812 links between disorders of the same class, an 8-fold enrichment with respect to 107 ± 10 links obtained between the same set of nodes in the randomized networks. This local functional clustering accounts for the small size of the giant components observed in the actual networks.

Disease-Associated Genes Identify Distinct Functional Modules. For several disorders known to arise from mutations in any one of a few distinct genes, the corresponding protein products have been shown to participate in the same cellular pathway, molecular complex, or functional module (21, 22). For example, Fanconi anemia arises from mutations in a set of genes encoding proteins involved in DNA repair, many of them forming a single heteromeric complex (23). Yet, the extent to which most disorders and disorder classes correspond to distinct functional modules in the cellular network has remained largely unclear. If genes linked by disorder associations encode proteins that interact in functionally distinguishable modules, then the proteins within such disease modules should more likely interact with one another than with other proteins. To test this hypothesis, we overlaid the DGN on a network of physical protein–protein interactions derived from high-quality systematic interactome mapping (6, 7) and literature curation (6). We found that 290 interactions overlap between the two networks, a 10-fold increase relative to random expectation ($P < 10^{-6}$; Fig. 3*a*).

Genes associated with the same disorder share common cellular and functional characteristics, as annotated in the Gene Ontology (GO) (24). If the HDN shows modular organization, then a group of genes associated with the same common disorder should share similar cellular and functional characteristics, as annotated in GO. To investigate the validity of this hypothesis, we measured the GO homogeneity of each disorder (see *SI Text*) separately for each branch of GO, biological process, molecular function, and cellular component, finding significant elevation of GO homogeneity with respect to random controls in all three branches (SI Fig. 8).

Disease genes encoding proteins that interact within common functional modules should tend to be expressed in the same tissue. To measure this, we introduced the tissue-homogeneity coefficient of a disorder, defined as the maximum fraction of genes among those belonging to a common disorder that are expressed in a specific tissue in a microarray data set obtained for 10,594 genes across 36 healthy tissues (25). We found that 68% of disorders exhibited almost perfect tissue-homogeneity (Fig. 3*b*), compared with 51% expected by chance ($P < 10^{-5}$).

Finally, disease genes that participate in a common functional module should also show high expression profiling correlation (26). The distribution of Pearson correlation coefficients (PCCs) for the coexpression profiles of pairs of genes associated with the same disorder was shifted toward higher values compared with that of a random control (Fig. 3*c*; $P < 10^{-6}$, χ^2 test). Similarly, the average PCC over all pairs of genes within a given disorder shows a significant shift from the random reference (Fig. 3*d*), with a small but clearly distinguishable peak in the distribution around PCC ≈ 0.75. This peak corresponds to ≈33 disorders with average PCC > 0.6 for which all genes are highly coexpressed in most tissues, including Heinz body anemia (PCC = 0.935), Bethlem myopathy (PCC = 0.835), and spherocytosis (PCC = 0.656).

In summary, genes that contribute to a common disorder (*i*) show an increased tendency for their products to interact with each other through protein–protein interactions, (*ii*) have a tendency to be expressed together in specific tissues, (*iii*) tend to display high coexpression levels, (*iv*) exhibit synchronized expression as a group, and (*v*) tend to share GO terms. Together, these findings support the hypothesis of a global functional relatedness for disease genes and their products and offer a network-based model for the diseasome. Cellular networks are modular, consisting of groups of highly interconnected proteins responsible for specific cellular functions (21, 22). A disorder then represents the perturbation or breakdown of a specific functional module caused by variation in one or more of the components producing recognizable developmental and/or physiological abnormalities.

This model offers a network-based explanation for the emergence of complex or polygenic disorders: a phenotype often correlates with the inability of a particular functional module to carry out its basic functions. For extended modules, many different combinations of perturbed genes could incapacitate the module, as a result of which mutations in different genes will appear to lead to the same phenotype. This correlation between disease and functional modules can also inform our understanding of cellular networks by helping us to identify which genes are involved in the same cellular function or network module (21, 22).

Centrality and Peripherality. An early indication of the connection between the structure of a cellular network and its functional properties was the finding that in *Saccharomyces cerevisiae* highly connected proteins or "hubs" are more likely encoded by essential genes (15, 16). This prompted a number of recent studies (27, 28) to formulate the hypothesis that human disease genes should also have a tendency to encode hubs. Yet, previous measurements found only a weak correlation between disease genes and hubs (29), resulting in an important mystery: what is the role, if any, of the cellular network in human diseases? Are disease genes more likely to encode hubs in the cellular network?

Our initial analysis appears to support the hypothesis that disease genes, given their impact on the organism, display a tendency to encode hubs in the interactome (27, 28), finding that disease related proteins have a 32% larger number of interactions (6, 7) with other proteins (average degree) than the nondisease proteins (see SI Fig 9) and that high-degree proteins are more likely to be encoded by genes associated with diseases than proteins with few interactions ($P = 1.6 \times 10^{-17}$; Fig. 4*a*). Next, we show, however, that despite this

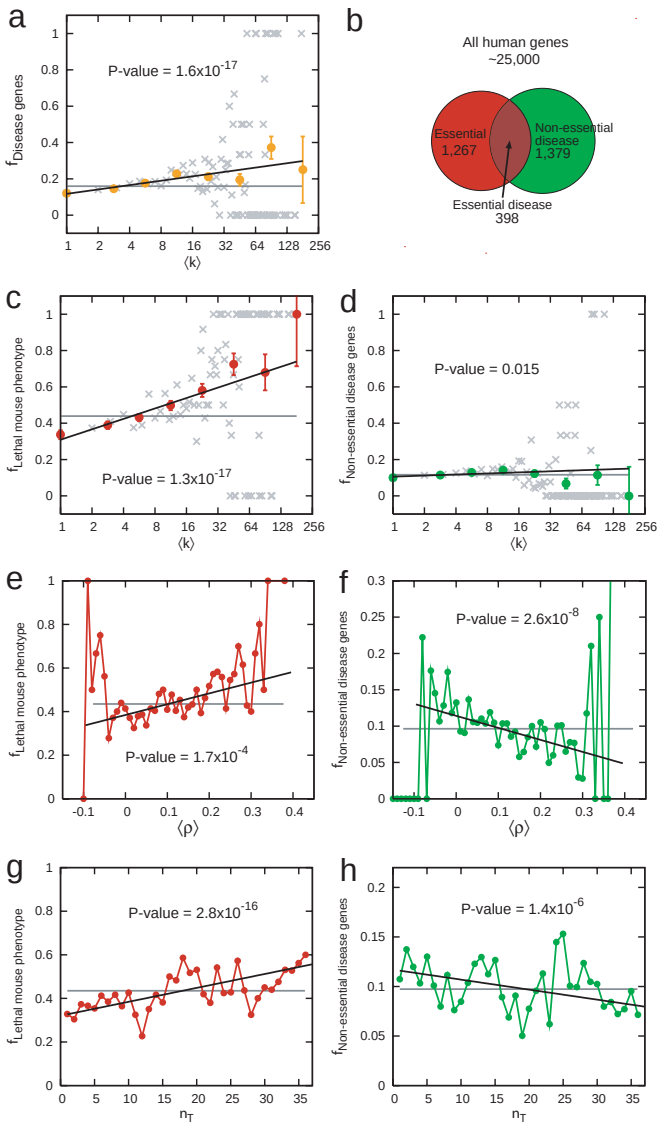

Fig. 4. Functional characteristics of disease and essential genes. (*a*) The fraction of disease genes among those whose protein products that interact with *k* other proteins. (*b*) Venn diagram showing the relationship between the human genes studied in this work. (*c*) The fraction of genes with lethal mouse phenotypes (essential genes) among those with mouse phenotypes that interact with *k* other proteins. (*d*) The same as in *a*, but only for nonessential disease genes, i.e., excluding 398 proteins with lethal mouse phenotypes. (*e* and *f*) The fraction of essential genes (*e*) and nonessential disease genes (*f*) among those whose average PCC with other genes is $\langle\rho\rangle$. (*g* and *h*) The fraction of essential genes (*g*) and nonessential disease genes (*h*) among those whose transcript is expressed in n_T tissues. Gray horizontal lines in *a* and *c*–*h* indicate the global average. Error bars represent standard errors. Note that for some data points the error bars are smaller than the symbol size, and thus are not visible. In *a*, *c*, and *d* gray symbols are the linearly binned data points, whereas color corresponds to the statistically more uniform log-binned data. For details of the significance analysis, see *SI Text*.

apparent correlation, the relationship between diseases and hubs hides deep differences between various disease genes.

When exploring whether disease genes encode hubs, we, and authors of other earlier studies (27–29), ignored the fact that some human genes are essential in early development and functional changes in these contribute to the high rate of first-trimester spontaneous abortions, which might be as much as 20% of recognized pregnancies. One strategy to explore the impact of this *in utero* essential segment of human disease is to consider human orthologs of mouse genes that result in embryonic or postnatal lethality when disrupted by homologous recombination (Mouse Genome Informatics; www.informatics.jax.org). All together, we find 1,267 such mouse lethal orthologs of human genes, of which 398 are associated with human diseases, representing 22% of all known human disease genes. This allows us to distinguish between two classes of human genes: 1,267 "essential genes" and 1,379 "nonessential disease genes," the latter obtained by removing from the full list of 1,777 OMIM disease genes the 398 that are also essential (Fig. 4*b*). Next, we show that these two classes of genes play quite different roles in the human interactome.

First, we find that essential proteins show a tendency to be associated with hubs ($P = 1.3 \times 10^{-17}$; Fig. 4*c*), displaying a much stronger trend than the one observed for all disease proteins (Fig. 4*a*). This raises an important question: Could the observed correlation between disease genes and hubs (Fig. 4*a*) be the sole consequence of the fact that a small fraction (22%) of disease genes is also essential? To address this question we measured the degree dependence of the nonessential disease proteins (Fig. 4*d*). Surprisingly, the correlation between hubs and disease proteins entirely disappears. Thus, the vast majority of disease genes (78%), those that are nonessential, do not show a tendency to encode hubs, indicating that the observed weak correlations between hubs and disease genes (Fig. 4*a*) was entirely due to the few essential genes within the disease gene class.

To carry on its basic functions, the cell needs to maintain the coordinated activity of important functional modules, driving in a relatively synchronized manner the expression patterns of the most important genes. Therefore, one expects that the expression pattern of both essential and disease genes will be synchronized with a significant number of other genes. To test this, we determined the average gene coexpression coefficient $\langle\rho\rangle_i = \Sigma_j \mathrm{PCC}_{ij}$ between an essential (or nonessential disease) gene *i* and all other genes in the cell, calculating the PCC_{ij} values from healthy human tissue microarray measurements (25). Confirming our expectation, for essential genes we find that genes that display high average coexpression $\langle\rho\rangle$ with all other genes are more likely to be essential than those that show small or negative $\langle\rho\rangle$ ($P = 1.7 \times 10^{-4}$; Fig. 4*e*). Surprisingly, however, nonessential disease genes show the opposite effect, being associated with genes whose expression pattern is anticorrelated or not-correlated with other genes, and underrepresented among the genes that are highly synchronized ($\langle\rho\rangle > 0.2$) ($P = 2.6 \times 10^{-8}$; Fig. 4*f*). Thus, the expression pattern of nonessential disease genes appears to be decoupled from the overall expression pattern of all other genes, whereas essential genes have a tendency to be coupled to the rest of the cell.

Finally, we asked whether housekeeping genes, expressed in all tissues, have a tendency to encode disease genes. We find that the more tissues in which a gene is expressed, the higher the likelihood that it will be essential ($P = 2.8 \times 10^{-16}$; Fig. 4*g*). The opposite is true for nonessential disease genes: they have a tendency to be expressed in a few tissues ($P = 1.4 \times 10^{-6}$; Fig. 4*h*). Similarly, we found that only 9.9% of housekeeping genes correspond to disease genes, compared with 13.5% of nonhousekeeping genes, a significant 36% difference ($P = 3.6 \times 10^{-6}$). In contrast, 59.8% of housekeeping genes annotated with mouse phenotype were essential, compared with 40.5% for nonhousekeeping genes ($P < 10^{-4}$).

These results support the somewhat unexpected conclusion that nonessential disease genes are not associated with hubs (27, 28), show smaller correlation in their expression pattern with the rest of the genes in the cell than expected from random, and have a tendency to be expressed in only a few tissues. Therefore, contrary to earlier hypotheses and our expectations, the vast majority of nonessential disease genes occupy functionally peripheral and topologically neutral positions in the cellular network. In stark contrast, essential genes are likely to encode hubs, show highly synchronized expression with the rest of the genes, and are expressed in most tissues, being overrepresented among housekeep-

ing genes. Thus, essential genes are topologically and functionally central.

This unexpected peripherality of most disease genes can be best explained by using an evolutionary argument. Mutations in topologically central, widely expressed genes are more likely to result in severe impairment of normal developmental and/or physiological function, leading to lethality *in utero* or early extrauterine life and to eventual deletion from the population. Only mutations compatible with survival into the reproductive years are likely to be maintained in a population. Therefore, disease-related mutations in the functionally and topologically peripheral regions of the cell give a higher chance of viability.

Disease genes whose mutations are somatic should not be subject to the selective pressure discussed above. Instead, somatic mutations that lead to severe disease phenotypes should more likely affect the functional center. To test the predictive power of this selection-based argument, we studied separately the properties of somatic cancer genes (Cancer Genome Census; www.sanger.ac.uk/genetics/CGP/Census) and found that they (*i*) are more likely to encode hubs, (*ii*) show higher coexpression with the rest of the genes in the cell, and (*iii*) are more represented among housekeeping genes (SI Fig. 10). The observed functional and topological centrality of somatic cancer genes fits well with our current understanding that many cancer genes play critical roles in cellular development and growth (11).

Discussion

Throughout history, clinicians and medical researchers have focused on a few disorder(s) sharing commonalities in etiology or pathology. Recent progress in genetics and genomics has led to an appreciation of the effects of gene mutations in virtually all disorders and provides the opportunity to study human diseases all at once rather than one at a time (4, 30). This unique approach offers the possibility of discerning general patterns and principles of human disease not readily apparent from the study of individual disorders.

An important tool in this quest is the HDN that represents a genome-wide roadmap for future studies on disease associations. The accompanying detailed diseasome map (SI Fig. 13), showing all disorders and the genes associated with different disorders, offers a rapid visual reference of the genetic links between disorders and disease genes, a valuable global perspective for physicians, genetic counselors, and biomedical researchers alike.

To test whether the conclusions obtained in this work are robust to the incompleteness of the OMIM coverage, we expanded our study to include not only genes with identified mutations linked to the specific disease phenotype, but also those that satisfy the less stringent criterion that the phenotype has not been mapped to a specific locus (18). This expansion increased the number of disease-associated genes from 1,777 to 2,765, but also introduced noise in the data, because the link between many of the newly added genes and diseases is less stringent. Yet, the overall organization of the expanded diseasome map remains largely unaltered (SI Fig. 11), and none of the trends uncovered in Fig. 4 are affected by this extension (SI Fig. 12), supporting the robustness of our findings to further expansion of the OMIM database. Thus, although the maps shown in Fig. 2 and SI Fig. 13 will inevitably undergo local changes with the discovery of new disease genes, this will not change the overall organization and layout of the HDN significantly, because the HDN reflects the underlying cellular network-based relationship between genes and functional modules.

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Supporting Information

Files in this Data Supplement:

SI Figure 5

Fig. 5. A subset of the Morbid Map, providing the disorder-disease gene associations. We parsed the MM list to merge eleven complementation groups of Fanconi anemia into a single disorder (id: 523) and two entries of fibromatosis into a single disorder (id: 537). The same procedure was reported for all entries, mapping 2,929 disorder entries in the MM into 1,286 distinct human disorder IDs. The full list of such disorders is provided in SI Table 1. The original MM list consists of the second to the fifth columns.

SI Figure 6

Fig. 6. Characterizing the topology of the HDN and the DGN. (*A* and *B*) Distribution of (*A*) the size *s* (number of genes involved in a disorder) and (*B*) the degree *k* (number of other disorders a disorder shares genes with) of all disorders in the HDN. Gray symbols correspond to a linear binning, while red dots represent the logarithmicallybinned data, maintaining the same statistical significance in each bin. The continuous lines represent the fit to the logbinned data, following the generalized powerlaw $f(x) = c(x + a)^b$ with (*A*) $b \approx 2.7$ and (*B*) $b \approx 6.5$, obtained from the least-square fit. (*C*) Distribution of the cluster sizes in the HDN (blue), where the isolated peak at 516 corresponds to the size of the giant component. The component size distribution for the randomized network is also shown (light blue). (*D*) Histogram of the number of disorders a gene is involved in, identifying the four genes associated with the largest number of disorders. (*E*) Distribution of the connected component sizes in the DGN (blue). The largest component contains 903 genes. The component size distribution for randomized networks is also shown (light blue).

SI Figure 7

Fig. 7. Genetic heterogeneity and connectivity of disorder classes. Red (blue) denotes the enrichment (depletion) of the measure in the corresponding cell. Dimcolored cell denotes the observed value is not statistically significant ($P > 10^2$). The statistical significance of each value is calculated with the randomized HDN obtained from the randomized disorderdisease gene associations.

SI Figure 8

Fig. 8. Gene Ontology Homogeneity. The GO homogeneity of disorders for the GO categories biological process (*Left*), molecular function (*Center*), and cellular component (*Right*). Red bars represent the actual histogram and the blue bars denote the random control, obtained for each disorder by choosing the same number of genes randomly.

SI Figure 9

Fig. 9. Average degrees of groups of human genes. Average degree of disease genes, essential genes,

and nonessential disease genes, as well as all genes. Error bar denotes the standard error.

SI Figure 10

Fig. 10. Centrality of somatic cancer genes. Plotted in each panel is the fraction of somatic cancer genes as a function of the average degree <*k*> (*Left*), the average coexpression coefficient <ρ> (*Center*), and the number of tissues expressed n_T (*Right*). All three quantities show positive trends, suggesting that somatic cancer genes are topologically and functionally central.

SI Figure 11

Fig. 11. Layout of the HDN with the extended data set. HDN with the extended dataset consists of 944 disorders with at least one link to other disorders and 576 disorders form the giant component.

SI Figure 12

Fig. 12. Functional characteristics of disease and essential genes for the extended data set. (*a*) The fraction of disease genes among those whose protein products interact with k other proteins. (*b*) The fraction of genes with lethal mouse phenotype among those whose protein products interact with k other proteins. (*c*) The same as in *a*, but excluding the proteins with lethal mouse phenotypes. (*d*) Average degree of disease, essential, and nonessential disease genes, as well as all genes (random). The fraction of essential genes (*e*) and disease genes (*f*) among those whose average PCC with other genes is <ρ>. Gray horizontal lines in *a*-c and e-f indicate the global average and the gray bars in *e-f* show the number of genes (without scales) in each bin indicating that the high fluctuations at low and high <ρ> are due to the small number of genes in those bins. (*g*) Average PCC of genes in different categories. (*h*) Fraction of disease genes (orange) and of genes with lethal (red) and nonlethal (light blue) mouse phenotypes among housekeeping and nonhousekeeping genes.

SI Figure 13

Fig. 13. Bipartite-graph representation of the diseasome. A disorder (circle) and a gene (rectangle) are connected if the gene is implicated in the disorder. The size of the circle represents the number of distinct genes associated with the disorder. Isolated disorders (disorders having no links to other disorders) are not shown. Also, only genes connecting disorders are shown.

SI Text

S1. Construction of the Diseasome Map

The most complete and best-curated list of known disorder-gene associations is maintained in the Morbid Map (MM) of the Online Mendelian Inheritance in Man (OMIM) (1). Each entry of the MM is composed of four fields, the name of the disorder, the associated gene symbols, its corresponding OMIM id, and the chromosomal location. We downloaded the MM file on December 21, 2005. Out of 4,043 MM entries, we selected 2,929 entries with the "(3)" tag, for which there is strong evidence that at least one mutation in the particular gene is causative to the disorder. We then parsed these 2,929 disorder terms into 1,284 distinct disorders by merging disease subtypes of a single disease, based on their given disorder names. For example, the eleven complementation groups of Fanconi anemia are merged into the disease "Fanconi anemia" and the two fibromatosis entries are merged into the disease "fibromatosis" [see Supporting Information (SI) Fig. 5]. The merging was done first automatically by running a string-match script and then each entry was verified manually. Each disease was then assigned a unique disease ID.

Subsequently we classified each disorder into 20 primary disorder classes manually, following the classification scheme shown in Fig. 2. The classification is based on the physiological system affected by the disorder. For example, 113 disorders constitute a "cancer" class and 41 disorders such as atherosclerosis and stroke constitute a "cardiovascular" class. When a disorder affects multiple systems we tried to assign it to a primary class based on which system was most affected; if a primary class was not evident then the disorder was placed into the "multiple" class. While disease categorization is often a cause of heated debate, occasional misclassification will not affect our results. Disorders having distinct multiple clinical features are assigned to the "multiple" class, with 155 disorders in this category. For 31 disorders there was insufficient information available for a clear class assignment, thus we annotated these into an "unclassified" class. Therefore every disorder was annotated into one of 22 disorder classes.

Finally, each gene symbol was mapped onto an Entrez ID, generating the list of disease gene-disorder class associations available as SI Table 1.

SI Tables 2 and 3 summarize the properties of the diseasome map. For each disorder Table 2 tabulates the disorder class, the degree *k* in the HDN, the class-degree κ (number of distinct disorder classes it connects to), the size *s* (number of associated genes) and the list of the associated disease genes. For each gene Table 3 tabulates the class of its associated disorders, and the number and the list of disorders to which it is associated.

S2. Properties of the HDN and DGN

The layouts of the giant components of both HDN and DGN in Fig. 2 were generated by a force-directed algorithm, followed by a local rearrangement for visual clarity, while leaving the network's overall layout unperturbed.

In the HDN, the number of genes associated with a disorder, *s*, has a broad distribution (SI Fig. 6*a*), indicating that most disorders relate to a few disease genes, while a handful of disorders relate to dozens of genes (SI Table 2). The HDN exhibits a skewed degree (*k*) distribution (SI Fig. 6*b*), with most disorders linked to only a few other disorders, while a few disorders (SI Table 2) represent hubs that are connected to a large number of distinct disorders.

In the complementary DGN, while the number of genes involved in multiple diseases decreases rapidly, several disease genes are involved in as many as ten disorders, representing major hubs in the network (SI Fig. 6*d*).

S3. Component Size Distributions of HDN and DGN

The topology of the HDN and GDN deviates from random. To obtain random controls we randomly shuffled the disorder-disease gene associations, while keeping both the number of genes that a disorder is associated with and the number of disorders that a gene is implicated in unchanged. From these randomized disorder-disease gene associations we obtained the two projections of randomized diseasome, the randomized HDN and the randomized GDN. To control for cluster size distribution, we generated 10^4 independent randomized samples.

The component size distributions, $n(s)$, defined as the number of components with size *s*, for the two network projections obtained after the randomizations are shown in light blue in SI Fig. 6 *c* and *e*. Both HDN and DGN have significantly smaller giant components than expected random, due to the functional clustering. Thus, HDN and DGN represent an intermediate structure between a completely randomized network with a very large giant component and a functionally fully segregated network which would be broken into isolated clusters, each representing a disorder class. Apart from the giant component, in both networks the sizes of disconnected components are distributed approximately as a power law, with the exponent approximately -3 both times. A solid conclusion on the distribution is difficult to draw due to the limited statistics.

S4. Genetic Heterogeneity and Connectivity of Disorder Classes

Genetic heterogeneity, specifically locus heterogeneity, means that mutations in more than one gene lead to similar disorder phenotypes (2). We measured the genetic heterogeneity of a disorder class as the average number of genes in the disorders belonging to the selected class (i.e., the average size of nodes in the class in the HDN). To quantify the statistical significance of the observed values, we randomized class annotations by randomly shuffling the disorder-class associations. According to the obtained *P*-values we identified significantly enriched (red) and significantly depleted (blue) classes (SI Fig. 7). The cancer and neurological disorder classes show high genetic heterogeneity, while the metabolic, skeletal, and multiple disorder classes show low genetic heterogeneity. We also calculated for each disorder class the fraction of disorders that are connected to each other in the HDN to quantify the "connectivity" of the particular disorder class. The statistical significance of the observed connectivity was assessed by the randomized disorder-disease gene associations (SI Fig. 7). By this measure, cancer is the most connected class and the metabolic disorder class is the least connected.

S5. Protein-Protein Interaction Data

To obtain a comprehensive human protein-protein interaction (PPI) data we combined two high quality systematic yeast two-hybrid experiments (3, 4) with PPIs obtained from literature by manual curation (3). The integrated set of PPIs contains 22,052 non-self-interacting, non-redundant interactions between 7,533 genes. The list of PPIs used is available as SI Table 4.

S6. Random Control for the PPI-GDN Overlap

To generate the random control of the overlap between the PPI network and the GDN in Fig. 3*a*, we first identified 1,203 genes that are present both in the PPI network and the GDN and for each disorder *i* the number of genes n_i that are present in the PPI network. To obtain the random control of the overlap, we calculated for each disease *i* the number of PPIs between the n_i nodes selected randomly among the 1,203 genes while keeping the degrees of the associated nodes. We performed this procedure for every disorder to obtain the number of all overlaps for a single random configuration. We generated 10^6 independent random configurations to obtain significant statistics and *P*-value.

S7. Gene Ontology Analysis

If the HDN shows modular organization then a group of genes associated with the same common disorder should share similar cellular and functional characteristics, as annotated in Gene Ontology (GO; ref. 5). To investigate this, we measured the GO homogeneity (GH) of each disorder as the maximum fraction of genes in the same disorder that have the same GO terms. It is defined as

$$GH_i = \max_j [n^j_i/n_i],$$

where in this case n_i denotes the number of genes in the disorder *i* that have any GO annotations, and

n^j_i the number of genes that have the specific GO term j. We calculated GH_i separately for each branch of GO, biological process (BP), molecular function (MF), and cellular component (CC). As expected, we find a significant elevation in the GH with respect to random controls in all three branches (SI Fig. 8). For example, we find a 23-fold increase in the perfect homogeneity for BP (79% vs. 3.4%), a 13-fold increase (75% vs. 5.5%) for MF, and a 9-fold increase (79% vs. 8.8%) for CC. To obtain the random control of the GO homogeneity distribution for each disorder we picked the same number of genes randomly in the GO annotation data and calculated their GO homogeneity. We generated 10^4 random instances to reach statistical significance.

S8. Gene Expression Microarray Data

To calculate the coexpression correlation between wild-type human gene transcripts, we used microarray data available for 36 normal human tissues (6). By matching Entrez gene ID, 1,357 human disease genes had corresponding microarray probes (76% of human disease genes). A gene is considered to be "expressed" if the P-value associated with its transcript abundance is less than the threshold, $P < 0.02$ (6). We consider a gene as housekeeping gene if it is expressed in all 36 tissues. Genes that are not expressed in any examined tissues are excluded from the analysis.

S9. Tissue Homogeneity

The tissue homogeneity (TH) coefficient quantifies whether genes that are implicated in the same disorders tend to be expressed in similar human tissues. We define the TH of a disorder i as

$$TH_i = \max_j [n^j_i/n_i],$$

where n_i denotes the number of genes in the disorder i that are expressed in at least one tissue, n^j_i the number of genes that are expressed in the tissue j among them, and $\max_j [\cdot]$ denotes the function returning the maximum-value argument across j. TH has the maximal value 1 if all of the genes are expressed together in at least one tissue, and takes the minimum value $1/n$ when all are expressed in different tissues. To obtain the random control of the tissue homogeneity distribution we picked the same number of genes randomly in the microarray data for each disorder and calculated their tissue homogeneity. We generated 10^5 random instances to reach statistical significance.

S10. Random Controls for Gene Expression Analysis

To obtain the random control of the Pearson correlation coefficient (PCC) distributions for the gene expression in Fig. 3*c* and *d*, we calculated the distribution of all gene pairs in the microarray data (Fig. 3*c*) and the average PCC between the same number of genes chosen randomly from the microarray data (Fig. 3*d*). To obtain the P-values, we perform the χ^2 -test, calculating χ^2 values between the random normalized histograms obtained from the reference distributions (blue) and the actual distribution (red), performing 10^6 independent runs to obtain significant statistics.

S11. Mouse Phenotype Data

To predict the essentiality of a human gene, we used the phenotype information of the corresponding mouse orthologs. A human gene was defined as "essential" if a knock-out of its mouse ortholog confers lethality. We obtained the human-mouse orthology and mouse phenotype data from Mouse Genome Informatics (http://www.informatics.jax.org) on January 3, 2006. We considered the classes of embryonic/prenatal lethality and postnatal lethality as lethal phenotypes, and the rest of phenotypes as non-lethal ones. There were 1,267 mouse-lethal human orthologs, of which 398 have known human disease associations (22% of human disease genes).

S12. Significance Analyses of the Results in Fig. 4

To assess the statistical significance of the reported results, we apply the linear regression model and performed the χ^2 test for the significance of the measured trends dictated by the regression coefficient (7). In particular, we take $\log_2 k$ as the dependent variable in Fig. 4*a*, *c*, and *d*, which is more appropriate than k due to the power-law degree distribution. We found that the measured trends described by the linear regression model are statistically significant, with associated with P values 4.2×10^{-6} (*a*), 2.8×10^{-5} (*c*), 1.4×10^{-4} (*d*), 1.1×10^{-16} (*e*), and 3.5×10^{-7} (*f*).

In addition, we assess the significance of the "strength" of the trends using the linear regression model. The slope A is obtained as the coefficient in the linear regression model and quantifies the strength (magnitude) of the trend. The P value of the observed trend A_O will be the probability that we have A equal or larger (smaller, for the negative trend in *f*) than A_O purely by chance. To calculate the P value, we randomized our sample by which we randomly redistribute the attributes (genes associated with diseases in *a*, for example) and perform the linear regression to obtain randomized (null) values of A, denoted by A_R. We found that A_R approximately follows a Gaussian distribution with zero mean and standard deviation σ_A. Thus, we can calculate the P value of the observed A_O from the Z-score defined as $Z = (A_O - \langle A_R \rangle)/\sigma_A = A_O/\sigma_A$. In SI Table 5, we summarize the results of the significance analysis according to the described procedure.

S13. Centrality of Somatic Cancer Genes

The selection-based argument presented in the paper does not apply to the diseases that are caused by somatic mutations. Thus, for example, cancers caused by somatic mutations need not be at the functional periphery. Instead, given the severe physiological damage, often leading to death, resulting from such mutations, these mutations are expected to affect the functional center of the cell. To test this, we studied the properties of genes whose somatic mutations are known to induce cancer. We obtained the list of somatic cancer genes from the Cancer Gene Census (www.sanger.ac.uk/genetics/CGP/Census). From the analysis (SI Fig. 10) we found that these somatic cancer genes indeed are (*i*) more likely to encode hub proteins, (*ii*) show higher coexpression with the rest of the genes in the cell,

and (*iii*) are more represented among housekeeping genes, confirming our expectation that somatic cancer genes are functionally central.

S14. Analysis of the Extended Data Set

In the analysis presented in the paper we included only the disorder-disease gene associations for which the wild-type gene is mapped and the mutation thereof is clearly demonstrated to be associated with the disorder, among all of the data catalogued in the Morbid Map in OMIM (1). In this section, we relax this criterion, and perform the same analysis with all of the disorder-disease genes associations listed in the Morbid Map, including those with weaker evidences, for which only the mapping of either the wild-type gene [tag "(1)"] or the disease phenotype itself [tag "(2)"] is known (1). Technically this extension did not require any further data curation, given that entries denoted by "(3)" tag in the OMIM database correspond to genes for which there is strong evidence that at least one mutation in the particular gene is causative of the disorder. In the earlier study we focused only on these genes (see Section S1). In the extended study presented here, we included also those entries that have "(1)" or "(2)" tags. This extension will increase the coverage of the data but at the same time introduce potential errors in the form of disease genes that may not turn out to be associated with the specific disorder. The objective of this extension is to test the robustness of the main findings of current study to the increase in the coverage and the presence of noise in the dataset.

With this extension, we obtain a list of associations between 1,578 disorders and 2,765 disease genes, from 4,043 Morbid Map entries as of December 21, 2005. Thus the coverage in the genome increases by 50%. First we generate the analog of Fig. 2*a*, the layout of the HDN (SI Fig. 11). The overall layout of the disorder classes and the overall position of diseases remain largely unaltered. Next we perform the analysis analogous to that shown in Fig. 4. The overall behavior that (*i*) the essential genes likely encode hubs whereas the nonessential disease genes are not particularly associated with hubs and (*ii*) the highly coexpressed genes are depleted with the disease genes while enriched with essential genes is also manifest clearly (SI Fig. 12). Overall, the main findings of the current study are robust to the extension of the dataset which both increases the coverage and introduces potential errors.

Supporting References

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Supporting Information Table 1. **Curated Morbid Map file with disease ID and class assignment (December 21, 2005 version).**					
Disease ID	Disorder name	Gene symbols	OMIM ID	Chromosome	Class
1	17,20-lyase deficiency, isolated, 202110 (3)	CYP17A1, CYP17, P450C17	609300	10q24.3	Endocrine
1	17-alpha-hydroxylase/17,20-lyase deficiency, 202110 (3)	CYP17A1, CYP17, P450C17	609300	10q24.3	Endocrine
3	2-methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency, 300438 (3)	HADH2, ERAB	300256	Xp11.2	Metabolic
4	2-methylbutyrylglycinuria (3)	ACADSB	600301	10q25-q26	Metabolic
5	3-beta-hydroxysteroid dehydrogenase, type II, deficiency (3)	HSD3B2	201810	1p13.1	Metabolic
6	3-hydroxyacyl-CoA dehydrogenase deficiency, 609609 (3)	HADHSC, SCHAD	601609	4q22-q26	Metabolic
7	3-Methylcrotonyl-CoA carboxylase 1 deficiency, 210200 (3)	MCCC1, MCCA	609010	3q25-q27	Metabolic
7	3-Methylcrotonyl-CoA carboxylase 2 deficiency, 210210 (3)	MCCC2, MCCB	609014	5q12-q13	Metabolic
8	3-methylglutaconic aciduria, type I, 250950 (3)	AUH	600529	Chr.9	Metabolic
9	3-methylglutaconicaciduria, type III, 258501 (3)	OPA3, MGA3	606580	19q13.2-q13.3	Metabolic
10	3-M syndrome, 273750 (3)	CUL7	609577	6p21.1	multiple
12	6-mercaptopurine sensitivity (3)	TPMT	187680	6p22.3	Metabolic
13	Aarskog-Scott syndrome (3)	FGD1, FGDY, AAS	305400	Xp11.21	multiple
14	Abacavir hypersensitivity, susceptibility to (3)	HLA-B	142830	6p21.3	Immunological
15	ABCD syndrome, 600501 (3)	EDNRB, HSCR2, ABCDS	131244	13q22	multiple
17	Abetalipoproteinemia, 200100 (3)	MTP	157147	4q22-q24	Metabolic
17	Abetalipoproteinemia (3)	APOB, FLDB	107730	2p24	Metabolic
18	Acampomelic campolelic dysplasia, 114290 (3)	SOX9, CMD1, SRA1	608160	17q24.3-q25.1	Skeletal
21	Acatalasemia (3)	CAT	115500	11p13	Hematological
22	Accelerated tumor formation, susceptibility to (3)	MDM2	164785	12q14.3-q15	Cancer
24	Achalasia-addisonianism-alacrimia syndrome, 231550 (3)	AAAS, AAA	605378	12q13	multiple
25	Acheiropody, 200500 (3)	C7orf2, ACHP, LMBR1	605522	7q36	Skeletal
26	Achondrogenesis-hypochondrogenesis, type II, 200610 (3)	COL2A1	120140	12q13.11-q13.2	Bone
27	Achondrogenesis Ib, 600972 (3)	SLC26A2, DTD, DTDST, D5S1708, EDM4	606718	5q32-q33.1	Bone
28	Achondroplasia, 100800 (3)	FGFR3, ACH	134934	4p16.3	Skeletal
29	Achromatopsia-2, 216900 (3)	CNGA3, CNG3, ACHM2	600053	2q11	Ophthamological
29	Achromatopsia-3, 262300 (3)	CNGB3, ACHM3	605080	8q21-q22	Ophthamological
29	Achromatopsia-4 (3)	GNAT2, ACHM4	139340	1p13	Ophthamological
30	Acid-labile subunit, deficiency of (3)	IGFALS, ALS	601489	16p13.3	Endocrine
31	Acquired long QT syndrome, susceptibility to (3)	KCNH2, LQT2, HERG	152427	7q35-q36	Cardiovascular
32	Acrocallosal syndrome, 200990 (3)	GLI3, PAPA, PAPB, ACLS	165240	7p13	multiple
33	Acrocapitofemoral dysplasia, 607778 (3)	IHH, BDA1	600726	2q33-q35	Skeletal
34	Acrodermatitis enteropathica, 201100 (3)	SLC39A4, ZIP4	607059	8q24.3	Dermatological
36	Acrokeratosis verruciformis, 101900 (3)	ATP2A2, ATP2B, DAR	108740	12q23-q24.1	Dermatological
38	Acromegaly, 102200 (3)	GNAS, GNAS1, GPSA, POH, PHP1B, PHP1A, AHO	139320	20q13.2	Endocrine
38	Acromegaly, 102200 (3)	SSTR5	182455	16p13.3	Endocrine
39	Acromesomelic dysplasia, Hunter-Thompson type, 201250 (3)	GDF5, CDMP1	601146	20q11.2	Skeletal
39	Acromesomelic dysplasia, Maroteaux type, 602875 (3)	NPR2, ANPRB, AMDM	108961	9p21-p12	Skeletal
44	Acyl-CoA dehydrogenase, long chain, deficiency of (3)	ACADL, LCAD	201460	2q34-q35	Metabolic
44	Acyl-CoA dehydrogenase, medium chain, deficiency of, 201450 (3)	ACADM, MCAD	607008	1p31	Metabolic
44	Acyl-CoA dehydrogenase, short-chain, deficiency of, 201470 (3)	ACADS, SCAD	606885	12q22-qter	Metabolic
45	Adenocarcinoma of lung, response to tyrosine kinase inhibitor in, 211980 (3)	EGFR	131550	7p12.3-p12.1	Cancer
45	Adenocarcinoma of lung, somatic, 211980 (3)	BRAF	164757	7q34	Cancer
45	Adenocarcinoma of lung, somatic, 211980 (3)	ERBB2, NGL, NEU, HER2	164870	17q21.1	Cancer
45	Adenocarcinoma of lung, somatic, 211980 (3)	PRKN, PARK2, PDJ	602544	6q25.2-q27	Cancer
45	Adenocarcinoma, ovarian, somatic (3)	PRKN, PARK2, PDJ	602544	6q25.2-q27	Cancer

46	Adenoma, periampullary (3)	APC, GS, FPC	175100	5q21-q22	Cancer
47	Adenomas, multiple colorectal, 608456 (3)	MUTYH	604933	1p34.3-p32.1	Cancer
47	Adenomas, salivary gland pleomorphic, 181030 (3)	PLAG1, SGPA, PSA	603026	8q12	Cancer
47	Adenomatous polyposis coli (3)	APC, GS, FPC	175100	5q21-q22	Cancer
47	Adenomatous polyposis coli, attenuated (3)	APC, GS, FPC	175100	5q21-q22	Cancer
49	Adenosine deaminase deficiency, partial, 102700 (3)	ADA	608958	20q13.11	Immunological
50	Adenylosuccinase deficiency, 103050 (3)	ADSL	608222	22q13.1	Metabolic
52	Adiponectin deficiency (3)	APM1, GBP28	605441	3q27	Endocrine
2053	Adrenal adenoma, sporadic (3)	MEN1	131100	11q13	Cancer
2054	Adrenal cortical carcinoma, 202300 (3)	TP53, P53, LFS1	191170	17p13.1	Cancer
53	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency (3)	CYP11B1, P450C11, FHI	202010	8q21	Endocrine
53	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency (3)	CYP21A2, CYP21, CA21H	201910	6p21.3	Endocrine
53	Adrenal hyperplasia, congenital, due to combined P450C17 and P450C21 deficiency, 201750 (3)	POR	124015	7q11.2	Endocrine
53	Adrenal hypoplasia, congenital, with hypogonadotropic hypogonadism, 300200 (3)	DAX1, AHC, AHX, NROB1	300473	Xp21.3-p21.2	Endocrine
2055	Adrenocortical insufficiency without ovarian defect (3)	FTZF1, FTZ1, SF1	184757	9q33	Endocrine
54	Adrenocortical tumor, somatic (3)	PRKAR1A, TSE1, CNC1, CAR	188830	17q23-q24	Cancer
55	Adrenocorticotropic hormone deficiency, 201400 (3)	TBS19	604614	1q23-q24	Endocrine
56	Adrenoleukodystrophy, 300100 (3)	ABCD1, ALD, AMN	300371	Xq28	Neurological
56	Adrenoleukodystrophy, neonatal, 202370 (3)	PEX10, NALD	602859	Chr.1	Neurological
56	Adrenoleukodystrophy, neonatal, 202370 (3)	PEX13, ZWS, NALD	601789	2p15	Neurological
56	Adrenoleukodystrophy, neonatal, 202370 (3)	PEX1, ZWS1	602136	7q21-q22	Neurological
56	Adrenoleukodystrophy, neonatal, 202370 (3)	PEX26	608666	22q11.21	Neurological
56	Adrenoleukodystrophy, neonatal, 202370 (3)	PXR1, PEX5, PTS1R	600414	12p13.3	Neurological
57	Adrenomyeloneuropathy, 300100 (3)	ABCD1, ALD, AMN	300371	Xq28	Metabolic
58	Adult i phenotype with congenital cataract, 110800 (3)	GCNT2	600429	6p24-p23	Hematological
58	Adult i phenotype without cataract, 110800 (3)	GCNT2	600429	6p24-p23	Hematological
59	ADULT syndrome, 103285 (3)	TP73L, TP63, KET, EEC3, SHFM4, LMS, RHS	603273	3q27	multiple
60	Advanced sleep phase syndrome, familial, 604348 (3)	PER2, FASPS, KIAA0347	603426	2q37.3	Neurological
61	Afibrinogenemia, 202400 (3)	FGA	134820	4q28	Hematological
61	Afibrinogenemia, congenital, 202400 (3)	FGB	134830	4q28	Hematological
63	Agammaglobulinemia, 601495 (3)	IGHM, MU	147020	14q32.33	Hematological
63	Agammaglobulinemia, autosomal recessive (3)	IGLL1, IGO, IGL5, VPREB2	146770	22q11.21	Hematological
63	Agammaglobulinemia, non-Bruton type, 601495 (3)	LRRC8, KIAA1437	608360	9q34.13	Hematological
63	Agammaglobulinemia, type 1, X-linked (3)	BTK, AGMX1, IMD1, XLA, AT	300300	Xq21.3-q22	Hematological
64	AGAT deficiency (3)	GATM, AGAT	602360	15q15.3	Metabolic
65	Agenesis of the corpus callosum with peripheral neuropathy, 218000 (3)	SLC12A6, KCC3A, KCC3B, KCC3, ACCPN	604878	15q13-q14	Neurological
69	AICA-ribosiduria due to ATIC deficiency, 608688 (3)	ATIC, PURH, AICAR	601731	2q35	Metabolic
70	AIDS, delayed/rapid progression to (3)	KIR3DL1, NKAT3, NKB1, AMB11, KIR3DS1	604946	19q13.4	Immunological
70	AIDS, rapid progression to, 609423 (3)	IFNG	147570	12q14	Immunological
70	AIDS, resistance to (3)	CXCL12, SDF1	600835	10q11.1	Immunological
71	Alagille syndrome, 118450 (3)	JAG1, AGS, AHD	601920	20p12	multiple
72	Albinism, brown oculocutaneous, (3)	OCA2, P, PED, D15S12, BOCA	203200	15q11.2-q12	Dermatological
72	Albinism, ocular, autosomal recessive (3)	OCA2, P, PED, D15S12, BOCA	203200	15q11.2-q12	Dermatological
72	Albinism, oculocutaneous, type IA, 203100 (3)	TYR	606933	11q14-q21	Dermatological
72	Albinism, oculocutaneous, type IB, 606952 (3)	TYR	606933	11q14-q21	Dermatological

72	Albinism, oculocutaneous, type II (3)	OCA2, P, PED, D15S12, BOCA	203200	15q11.2-q12	Dermatological
72	Albinism, rufous, 278400 (3)	TYRP1, CAS2, GP75	115501	9p23	Dermatological
74	Alcohol dependence, susceptibility to, 103780 (3)	HTR2A	182135	13q14-q21	Unclassified
2074	Alcohol intolerance, acute (3)	ALDH2	100650	12q24.2	Metabolic
75	Alcoholism, susceptibility to, 103780 (3)	GABRA2	137140	4p13-p12	Unclassified
76	Aldolase A deficiency (3)	ALDOA	103850	16q22-q24	Metabolic
77	Aldosterone to renin ratio raised (3)	CYP11B2	124080	8q21	Endocrine
78	Aldosteronism, glucocorticoid-remediable, 103900 (3)	CYP11B1, P450C11, FHI	202010	8q21	Endocrine
79	Alexander disease, 203450 (3)	GFAP	137780	17q21	multiple
79	Alexander disease, 203450 (3)	NDUFV1, UQOR1	161015	11q13	multiple
80	Alkaptonuria, 203500 (3)	HGD, AKU	607474	3q21-q23	Metabolic
82	Allan-Herndon-Dudley syndrome, 300523 (3)	SLC16A2, DXS128, XPCT	300095	Xq13.2	Neurological
83	Allergic rhinitis, susceptibility to, 607154 (3)	IL13, ALRH	147683	5q31	Immunological
85	Alopecia universalis, 203655 (3)	HR, AU	602302	8p21.2	Dermatological
86	Alpers syndrome, 203700 (3)	POLG, POLG1, POLGA, PEO	174763	15q25	Neurological
87	Alpha-1-antichymotrypsin deficiency (3)	SERPINA3, AACT, ACT	107280	14q32.1	Respiratory
88	Alpha-actinin-3 deficiency (3)	ACTN3	102574	11q13-q14	Muscular
90	Alpha-methylacetoacetic aciduria, 203750 (3)	ACAT1	607809	11q22.3-q23.1	Metabolic
91	Alpha-methylacyl-CoA racemase deficiency (3)	AMACR	604489	5p13.2-q11.1	Metabolic
92	Alpha-thalassemia/mental retardation syndrome, 301040 (3)	ATRX, XH2, XNP, MRXS3, SHS	300032	Xq13	Hematological
92	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 (3)	ATRX, XH2, XNP, MRXS3, SHS	300032	Xq13	Hematological
94	Alport syndrome, 301050 (3)	COL4A5, ATS, ASLN	303630	Xq22.3	Renal
94	Alport syndrome, autosomal recessive, 203780 (3)	COL4A3	120070	2q36-q37	Renal
94	Alport syndrome, autosomal recessive, 203780 (3)	COL4A4	120131	2q36-q37	Renal
95	Alstrom syndrome, 203800 (3)	ALMS1, ALSS, KIAA0328	606844	2p13	Neurological
96	Alternating hemiplegia of childhood, 104290 (3)	ATP1A2, FHM2, MHP2	182340	1q21-q23	Neurological
97	Alveolar soft-part sarcoma, 606243 (3)	ASPCR1, RCC17, ASPL, ASPS	606236	17q25	Cancer
98	Alzheimer disease-1, APP-related (3)	APP, AAA, CVAP, AD1	104760	21q21	Neurological
98	Alzheimer disease-2, 104310 (3)	APOE, AD2	107741	19q13.2	Neurological
98	Alzheimer disease-4, 606889 (3)	PSEN2, AD4, STM2	600759	1q31-q42	Neurological
98	Alzheimer disease, late-onset, 104300 (3)	APBB2, FE65L1	602710	4p14	Neurological
98	Alzheimer disease, late-onset, susceptibility to, 104300 (3)	NOS3	163729	7q36	Neurological
98	Alzheimer disease, late-onset, susceptibility to, 104300 (3)	PLAU, URK	191840	10q24	Neurological
98	Alzheimer disease, susceptibility to, 104300 (3)	ACE, DCP1, ACE1	106180	17q23	Neurological
98	Alzheimer disease, susceptibility to, 104300 (3)	MPO	606989	17q23.1	Neurological
98	Alzheimer disease, susceptibility to, 104300 (3)	PACIP1, PAXIP1L, PTIP	608254	7q36	Neurological
98	Alzheimer disease, susceptibility to (3)	A2M	103950	12p13.3-p12.3	Neurological
98	Alzheimer disease, susceptibility to (3)	BLMH, BMH	602403	17q11.2	Neurological
98	Alzheimer disease, type 3, 607822 (3)	PSEN1, AD3	104311	14q24.3	Neurological
98	Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822 (3)	PSEN1, AD3	104311	14q24.3	Neurological
98	Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822 (3)	PSEN1, AD3	104311	14q24.3	Neurological
99	Amelogenesis imperfecta 2, hypoplastic local, 104500 (3)	ENAM	606585	4q21	Bone
99	Amelogenesis imperfecta, 301200 (3)	AMELX, AMG, AIH1, AMGX	300391	Xp22.3-p22.1	Bone
99	Amelogenesis imperfecta, hypomaturation-hypoplastic type, with taurodontism, 104510 (3)	DLX3, TDO	600525	17q21.3-q22	Bone
99	Amelogenesis imperfecta, hypoplastic, and openbite malocclusion, 608563 (3)	ENAM	606585	4q21	Bone
99	Amelogenesis imperfecta, pigmented hypomaturation type, 204700 (3)	KLK4, EMSP1, PRSS17	603767	19q13.3-q13.4	Bone
100	Amish infantile epilepsy syndrome, 609056 (3)	SIAT9, ST3GALV	604402	2p11.2	Neurological
101	AMP deaminase deficiency, erythrocytic (3)	AMPD3	102772	11pter-p13	Hematological

102	Amyloid neuropathy, familial, several allelic types (3)	TTR, PALB	176300	18q11.2-q12.1	Neurological
103	Amyloidosis, 3 or more types (3)	APOA1	107680	11q23	Neurological
103	Amyloidosis, cerebroarterial, Dutch type (3)	APP, AAA, CVAP, AD1	104760	21q21	Neurological
103	Amyloidosis, Finnish type, 105120 (3)	GSN	137350	9q34	Neurological
103	Amyloidosis, hereditary renal, 105200 (3)	FGA	134820	4q28	Neurological
103	Amyloidosis, renal, 105200 (3)	LYZ	153450	Chr.12	Neurological
103	Amyloidosis, senile systemic (3)	TTR, PALB	176300	18q11.2-q12.1	Neurological
104	Amyotrophic lateral sclerosis 8, 608627 (3)	VAPB, VAPC, ALS8	605704	20q13.3	Neurological
104	Amyotrophic lateral sclerosis, due to SOD1 deficiency, 105400 (3)	SOD1, ALS1	147450	21q22.1	Neurological
104	Amyotrophic lateral sclerosis, juvenile, 205100 (3)	ALS2, ALSJ, PLSJ, IAHSP	606352	2q33	Neurological
104	Amyotrophic lateral sclerosis, susceptibility to, 105400 (3)	DCTN1	601143	2p13	Neurological
104	Amyotrophic lateral sclerosis, susceptibility to, 105400 (3)	NEFH	162230	22q12.2	Neurological
104	Amyotrophic lateral sclerosis, susceptibility to, 105400 (3)	PRPH	170710	12q12-q13	Neurological
105	Analbuminemia (3)	ALB	103600	4q11-q13	Hematological
107	Analgesia from kappa-opioid receptor agonist, female-specific (3)	MC1R	155555	16q24.3	Unclassified
108	Anderson disease, 607689 (3)	SARA2, SAR1B, CMRD	607690	5q31.1	Gastrointestinal
109	Androgen insensitivity, 300068 (3)	AR, DHTR, TFM, SBMA, KD, SMAX1	313700	Xq11-q12	Endocrine
110	Anemia, congenital dyserythropoietic, type I, 224120 (3)	CDAN1, CDA1	607465	15q15	Hematological
110	Anemia, Diamond-Blackfan, 105650 (3)	RPS19, DBA	603474	19q13.2	Hematological
110	Anemia, hemolytic, due to PK deficiency (3)	PKLR, PK1	266200	1q21	Hematological
110	Anemia, hemolytic, due to UMPH1 deficiency, 266120 (3)	NT5C3, UMPH1, PSN1	606224	7p15-p14	Hematological
110	Anemia, hemolytic, Rh-null, regulator type, 268150 (3)	RHAG, RH50A	180297	6p21.1-p11	Hematological
110	Anemia, hypochromic microcytic, 206100 (3)	NRAMP2	600523	12q13	Hematological
110	Anemia, neonatal hemolytic, fatal and near-fatal (3)	SPTB	182870	14q22-q23.2	Hematological
110	Anemia, sideroblastic/hypochromic (3)	ALAS2, ANH1, ASB	301300	Xp11.21	Hematological
110	Anemia, sideroblastic, with ataxia, 301310 (3)	ABCB7, ABC7, ASAT	300135	Xq13.1-q13.3	Hematological
2112	Aneurysm, familial arterial (3)	COL3A1	120180	2q31	Unclassified
113	Angelman syndrome, 105830 (3)	MECP2, RTT, PPMX, MRX16, MRX79	300005	Xq28	Developmental
113	Angelman syndrome, 105830 (3)	UBE3A, ANCR	601623	15q11-q13	Developmental
114	Angioedema, hereditary, 106100 (3)	C1NH, HAE1, HAE2, SERPING1	606860	11q11-q13.1	Immunological
114	Angioedema induced by ACE inhibitors, susceptibility to (3)	XPNPEP2	300145	Xq25	Immunological
115	Angiofibroma, sporadic (3)	MEN1	131100	11q13	Endocrine
117	Angiotensin I-converting enzyme, benign serum increase (3)	ACE, DCP1, ACE1	106180	17q23	Endocrine
118	Anhaptoglobinemia (3)	HP	140100	16q22.1	Hematological
119	Aniridia, type II, 106210 (3)	PAX6, AN2, MGDA	607108	11p13	Ophthamological
121	Ankylosing spoldylitis, susceptibility to, 106300 (3)	HLA-B	142830	6p21.3	Connective tissue
122	Anophthalmia 3, 206900 (3)	SOX2, ANOP3	184429	3q26.3-q27	Ophthamological
124	Anorexia nervosa, susceptibility to, 606788 (3)	HTR2A	182135	13q14-q21	Nutritional
126	Anterior segment anomalies and cataract (3)	EYA1, BOR	601653	8q13.3	Ophthamological
126	Anterior segment mesenchymal dysgenesis, 107250 (3)	FOXE3, FKHL12, ASMD	601094	1p32	Ophthamological
126	Anterior segment mesenchymal dysgenesis (3)	FOXC1, FKHL7, FREAC3	601090	6p25	Ophthamological
126	Anterior segment mesenchymal dysgenesis and cataract, 107250 (3)	PITX3	602669	10q25	Ophthamological
127	Antithrombin III deficiency (3)	AT3	107300	1q23-q25	Hematological
128	Antley-Bixler syndrome, 207410 (3)	POR	124015	7q11.2	Unclassified
129	Anxiety-related personality traits (3)	SLC6A4, HTT, OCD1	182138	17q11.1-q12	Psychiatric
130	Aortic aneurysm, ascending, and dissection (3)	FBN1, MFS1, WMS	134797	15q21.1	Cardiovascular
131	Apert syndrome, 101200 (3)	FGFR2, BEK, CFD1, JWS	176943	10q26	Connective tissue
132	Aplasia of lacrimal and salivary glands, 180920 (3)	FGF10	602115	5p13-p12	Gastrointestinal
133	Aplastic anemia, 609135 (3)	IFNG	147570	12q14	Hematological
133	Aplastic anemia, 609135 (3)	TERC, TRC3, TR	602322	3q21-q28	Hematological

133	Aplastic anemia, susceptibility to, 609135 (3)	TERT, TCS1, EST2	187270	5p15.33	Hematological
134	Apnea, postanesthetic (3)	BCHE, CHE1	177400	3q26.1-q26.2	Unclassified
136	ApoA-I and apoC-III deficiency, combined (3)	APOA1	107680	11q23	Metabolic
136	Apolipoprotein A-II deficiency (3)	APOA2	107670	1q21-q23	Metabolic
136	Apolipoprotein C3 deficiency (3)	APOC3	107720	11q23	Metabolic
136	Apolipoprotein H deficiency (3)	APOH	138700	17q23-qter	Metabolic
137	Apparent mineralocorticoid excess, hypertension due to (3)	HSD11B2, HSD11K	218030	16q22	Bone
138	Aquaporin-1 deficiency (3)	AQP1, CHIP28, CO	107776	7p14	Unclassified
139	ARC syndrome, 208085 (3)	VPS33B	608552	15q26.1	Gastrointestinal
140	Argininemia, 207800 (3)	ARG1	608313	6q23	Metabolic
140	Argininosuccinic aciduria, 207900 (3)	ASL	608310	7cen-q11.2	Metabolic
142	Aromatase deficiency (3)	CYP19A1, CYP19, ARO	107910	15q21.1	Metabolic
143	Aromatic L-amino acid decarboxylase deficiency, 608643 (3)	DDC	107930	7p11	Metabolic
144	Arrhythmogenic right ventricular dysplasia 2, 600996 (3)	RYR2, VTSIP	180902	1q42.1-q43	Cardiovascular
144	Arrhythmogenic right ventricular dysplasia 8, 607450 (3)	DSP, KPPS2, PPKS2	125647	6p24	Cardiovascular
144	Arrhythmogenic right ventricular dysplasia, familial, 9, 609040 (3)	PKP2, ARVD9	602861	12p11	Cardiovascular
146	Arthrogryposis multiplex congenita, distal, type 1, 108120 (3)	TPM2, TMSB, AMCD1, DA1	190990	9p13.2-p13.1	Developmental
146	Arthrogryposis multiplex congenita, distal, type 2B, 601680 (3)	TNNI2, AMCD2B, DA2B, FSSV	191043	11p15.5	Developmental
147	Arthropathy, progressive pseudorheumatoid, of childhood, 208230 (3)	WISP3, PPAC, PPD	603400	6q22-q23	Bone
146	Arthyrgryposis multiplex congenita, distal, type 2B, 601680 (3)	TNNT3, AMCD2B, DA2B, FSSV	600692	11p15.5	Developmental
150	Aspartylglucosaminuria (3)	AGA	208400	4q32-q33	Metabolic
151	Asperger syndrome, 300494 (3)	NLGN3	300336	Xq13	Psychiatric
151	Asperger syndrome, 300497 (3)	NLGN4, KIAA1260, AUTSX2	300427	Xp22.33	Psychiatric
153	Asthma, 600807 (3)	PHF11, NYREN34	607796	13q14.1	Respiratory
153	Asthma, atopic, susceptibility to (3)	MS4A2, FCER1B	147138	11q13	Respiratory
153	Asthma, dimished response to antileukotriene treatment in, 600807 (3)	ALOX5	152390	10q11.2	Respiratory
153	Asthma, nocturnal, susceptibility to (3)	ADRB2	109690	5q32-q34	Respiratory
153	Asthma, susceptibility to, 1, 607277 (3)	PTGDR, AS1	604687	14q22.1	Respiratory
153	Asthma, susceptibility to, 2, 608584 (3)	GPR154, GPRA, VRR1, PGR14	608595	7p15-p14	Respiratory
153	Asthma, susceptibility to (3)	HNMT	605238	1p32	Respiratory
153	Asthma, susceptibility to, 600807 (3)	IL12B, NKSF2	161561	5q31.1-q33.1	Respiratory
153	Asthma, susceptibility to, 600807 (3)	IL13, ALRH	147683	5q31	Respiratory
153	Asthma, susceptibility to, 600807 (3)	PLA2G7, PAFAH	601690	6p21.2-p12	Respiratory
153	Asthma, susceptibility to, 600807 (3)	SCGB3A2, UGRP1	606531	5q31-q34	Respiratory
153	Asthma, susceptibility to, 600807 (3)	TNF, TNFA	191160	6p21.3	Respiratory
153	Asthma, susceptibility to, 600807 (3)	UGB, CC10, CCSP, SCGB1A1	192020	11q12.3-q13.1	Respiratory
154	Ataxia, cerebellar, Cayman type, 601238 (3)	ATCAY, CLAC, KIAA1872	608179	19p13.3	Neurological
154	Ataxia, early-onset, with oculomotor apraxia and hypoalbuminemia, 208920 (3)	APTX, AOA, AOA1	606350	9p13.3	Neurological
154	Ataxia, episodic (3)	CACNB4, EJM	601949	2q22-q23	Neurological
154	Ataxia-ocular apraxia-2, 606002 (3)	SETX, SCAR1, AOA2	608465	9q34	Neurological
157	Ataxia-telangiectasia, 208900 (3)	ATM, ATA, AT1	607585	11q22.3	Immunological
157	Ataxia-telangiectasia-like disorder, 604391 (3)	MRE11A, MRE11, ATLD	600814	11q21	Immunological
154	Ataxia with isolated vitamin E deficiency, 277460 (3)	TTPA, TTP1, AVED	600415	8q13.1-q13.3	Neurological
160	Atelosteogenesis II, 256050 (3)	SLC26A2, DTD, DTDST, D5S1708, EDM4	606718	5q32-q33.1	Connective tissue
160	Atelostogenesis, type I, 108720 (3)	FLNB, SCT, AOI	603381	3p14.3	Connective tissue
162	Athabaskan brainstem dysgenesis syndrome, 601536 (3)	HOXA1, HOX1F, BSAS	142955	7p15.3	Neurological
163	Atherosclerosis, susceptibility to (3)	ALOX5	152390	10q11.2	Cardiovascular
164	Atopy, 147050 (3)	SPINK5, LEKTI	605010	5q32	Immunological
164	Atopy, resistance to, 147050 (3)	HAVCR1, HAVCR	606518	5q33.2	Immunological

164	Atopy, susceptibility to, 147050 (3)	PLA2G7, PAFAH	601690	6p21.2-p12	Immunological
164	Atopy, susceptibility to, 147050 (3)	SELP, GRMP	173610	1q23-q25	Immunological
164	Atopy, susceptibility to (3)	IL4R, IL4RA	147781	16p12.1-p11.2	Immunological
165	Atransferrinemia, 209300 (3)	TF	190000	3q21	Hematological
166	Atrial fibrillation, familial, 607554 (3)	KCNE2, MIRP1, LQT6	603796	21q22.1	Cardiovascular
166	Atrial fibrillation, familial, 607554 (3)	KCNQ1, KCNA9, LQT1, KVLQT1, ATFB1	607542	11p15.5	Cardiovascular
166	Atrial septal defect-2, 607941 (3)	GATA4	600576	8p23.1-p22	Cardiovascular
166	Atrial septal defect 3 (3)	MYH6, ASD3, MYHCA	160710	14q12	Cardiovascular
166	Atrial septal defect with atrioventricular conduction defects, 108900 (3)	NKX2E, CSX	600584	5q34	Cardiovascular
167	Atrichia with papular lesions, 209500 (3)	HR, AU	602302	8p21.2	Dermatological
168	Atrioventricular block, idiopathic second-degree (3)	NKX2E, CSX	600584	5q34	Cardiovascular
168	Atrioventricular septal defect, 600309 (3)	GJA1, CX43, ODDD, SDTY3, ODOD	121014	6q21-q23.2	Cardiovascular
168	Atrioventricular septal defect, partial, with heterotaxy syndrome, 606217 (3)	CRELD1, AVSD2	607170	3p25.3	Cardiovascular
168	Atrioventricular septal defect, susceptibility to, 2, 606217 (3)	CRELD1, AVSD2	607170	3p25.3	Cardiovascular
171	Attention deficit-hyperactivity disorder, susceptibility to, 143465 (3)	DRD5, DRD1B, DRD1L2	126453	4p16.1-p15.3	Psychiatric
173	Autism, susceptibility to, 209850 (3)	GLO1	138750	6p21.3-p21.2	Psychiatric
173	Autism, X-linked, 300425 (3)	MECP2, RTT, PPMX, MRX16, MRX79	300005	Xq28	Psychiatric
173	Autism, X-linked, 300425 (3)	NLGN3	300336	Xq13	Psychiatric
173	Autism, X-linked, 300495 (3)	NLGN4, KIAA1260, AUTSX2	300427	Xp22.33	Psychiatric
174	Autoimmune lymphoproliferative syndrome, 601859 (3)	TNFRSF6, APT1, FAS, CD95, ALPS1A	134637	10q24.1	Immunological
174	Autoimmune lymphoproliferative syndrome, type IA, 601859 (3)	TNFRSF6, APT1, FAS, CD95, ALPS1A	134637	10q24.1	Immunological
174	Autoimmune lymphoproliferative syndrome, type II, 603909 (3)	CASP10, MCH4, ALPS2	601762	2q33-q34	Immunological
174	Autoimmune lymphoproliferative syndrome, type IIB, 607271 (3)	CASP8, MCH5	601763	2q33-q34	Immunological
174	Autoimmune polyglandular disease, type I, 240300 (3)	AIRE, APECED	607358	21q22.3	Immunological
2174	Autoimmune thyroid disease, susceptibility to 3, 608175 (3)	TG, AITD3	188450	8q24.2-q24.3	Endocrine
175	Autonomic nervous system dysfunction (3)	DRD4	126452	11p15.5	Psychiatric
177	Axenfeld anomaly (3)	FOXC1, FKHL7, FREAC3	601090	6p25	Ophthamological
178	Azoospermia (3)	USP9Y, DFFRY	400005	Yq11.2	Endocrine
178	Azoospermia due to perturbations of meiosis, 270960 (3)	SYCP3, SCP3, COR1	604759	12q23	Endocrine
180	Bamforth-Lazarus syndrome, 241850 (3)	FOXE1, FKHL15, TITF2, TTF2	602617	9q22	Endocrine
182	Bannayan-Riley-Ruvalcaba syndrome, 153480 (3)	PTEN, MMAC1	601728	10q23.31	Unclassified
182	Bannayan-Zonana syndrome, 153480 (3)	PTEN, MMAC1	601728	10q23.31	Unclassified
183	Bardet-Biedl syndrome 1, 209900 (3)	BBS1	209901	11q13	multiple
183	Bardet-Biedl syndrome 1, modifier of, 209900 (3)	ARL6, BBS3	608845	3p12-q13	multiple
183	Bardet-Biedl syndrome, 209900 (3)	BBS7	607590	4q27	multiple
183	Bardet-Biedl syndrome 2, 209900 (3)	BBS2	606151	16q21	multiple
183	Bardet-Biedl syndrome 3, 600151 (3)	ARL6, BBS3	608845	3p12-q13	multiple
183	Bardet-Biedl syndrome 4, 209900 (3)	BBS4	600374	15q22.3-q23	multiple
183	Bardet-Biedl syndrome 5, 209900 (3)	BBS5	603650	2q31	multiple
183	Bardet-Biedl syndrome 6, 209900 (3)	MKKS, HMCS, KMS, MKS, BBS6	604896	20p12	multiple
183	Bardet-Biedl syndrome 8, 209900 (3)	TTC8, BBS8	608132	14q32.1	multiple
184	Bare lymphocyte syndrome, type I, 604571 (3)	TAPBP, TPSN	601962	6p21.3	Immunological
184	Bare lymphocyte syndrome, type I, due to TAP2 deficiency, 604571 (3)	TAP2, ABCB3, PSF2, RING11	170261	6p21.3	Immunological
184	Bare lymphocyte syndrome, type II, complementation group A, 209920 (3)	MHC2TA, C2TA	600005	16p13	Immunological
184	Bare lymphocyte syndrome, type II, complementation group C, 209920 (3)	RFX5	601863	1q21.1-q21.3	Immunological
184	Bare lymphocyte syndrome, type II, complementation group D, 209920 (3)	RFXAP	601861	13q14	Immunological
184	Bare lymphocyte syndrome, type II, complementation group E, 209920 (3)	RFX5	601863	1q21.1-q21.3	Immunological
185	Barth syndrome, 302060 (3)	TAZ, EFE2, BTHS, CMD3A, LVNCX	300394	Xq28	multiple

186	Bart-Pumphrey syndrome, 149200 (3)	GJB2, CX26, DFNB1, PPK, DFNA3, KID, HID	121011	13q11-q12	multiple
187	Bartter syndrome, type 1, 601678 (3)	SLC12A1, NKCC2	600839	15q15-q21.1	multiple
187	Bartter syndrome, type 2, 241200 (3)	KCNJ1, ROMK1	600359	11q24	multiple
187	Bartter syndrome, type 3, 607364 (3)	CLCNKB	602023	1p36	multiple
187	Bartter syndrome, type 4, 602522 (3)	BSND	606412	1p31	multiple
187	Bartter syndrome, type 4, digenic, 602522 (3)	CLCNKA	602024	1p36	multiple
187	Bartter syndrome, type 4, digenic, 602522 (3)	CLCNKB	602023	1p36	multiple
188	Basal cell carcinoma (3)	RASA1, GAP, CMAVM, PKWS	139150	5q13.3	Cancer
188	Basal cell carcinoma, somatic, 605462 (3)	PTCH2	603673	1p32	Cancer
188	Basal cell carcinoma, somatic, 605462 (3)	PTCH, NBCCS, BCNS, HPE7	601309	9q22.3	Cancer
188	Basal cell carcinoma, sporadic (3)	SMOH, SMO	601500	7q31-q32	Cancer
2188	Basal cell nevus syndrome, 109400 (3)	PTCH, NBCCS, BCNS, HPE7	601309	9q22.3	multiple
4188	Basal ganglia disease, adult-onset, 606159 (3)	FTL	134790	19q13.3-q13.4	Neurological
4188	Basal ganglia disease, biotin-responsive, 607483 (3)	SLC19A3	606152	2q36.3	Neurological
190	B-cell non-Hodgkin lymphoma, high-grade (3)	BCL7A, BCL7	601406	12q24.1	Cancer
191	BCG infection, generalized familial (3)	IFNGR1	107470	6q23-q24	Immunological
192	Beare-Stevenson cutis gyrata syndrome, 123790 (3)	FGFR2, BEK, CFD1, JWS	176943	10q26	multiple
193	Becker muscular dystrophy, 300376 (3)	DMD, BMD	300377	Xp21.2	Muscular
193	Becker muscular dystrophy modifier, 310200 (3)	MYF6	159991	12q21	Muscular
194	Beckwith-Wiedemann syndrome, 130650 (3)	CDKN1C, KIP2, BWS	600856	11p15.5	multiple
194	Beckwith-Wiedemann syndrome, 130650 (3)	H19, D11S813E, ASM1, BWS	103280	11p15.5	multiple
194	Beckwith-Wiedemann syndrome, 130650 (3)	KCNQ10T1, LIT1	604115	11p15.5	multiple
194	Beckwith-Wiedemann syndrome, 130650 (3)	NSD1, ARA267, STO	606681	5q35	multiple
195	Benzene toxicity, susceptibility to (3)	NQO1, DIA4, NMOR1	125860	16q22.1	Unclassified
196	Bernard-Soulier syndrome, 231200 (3)	GP1BA	606672	17pter-p12	Hematological
196	Bernard-Soulier syndrome, type B, 231200 (3)	GP1BB	138720	22q11.2	Hematological
196	Bernard-Soulier syndrome, type C (3)	GP9	173515	Chr.3	Hematological
197	Beryllium disease, chronic, susceptibility to (3)	HLA-DPB1	142858	6p21.3	Unclassified
198	Beta-2-adrenoreceptor agonist, reduced response to (3)	ADRB2	109690	5q32-q34	Unclassified
199	Beta-ureidopropionase deficiency (3)	UPB1, BUP1	606673	22q11.2	Metabolic
200	Bethlem myopathy, 158810 (3)	COL6A1, OPLL	120220	21q22.3	Muscular
200	Bethlem myopathy, 158810 (3)	COL6A2	120240	21q22.3	Muscular
200	Bethlem myopathy, 158810 (3)	COL6A3	120250	2q37	Muscular
201	Bietti crystalline corneoretinal dystrophy, 210370 (3)	CYP4V2, BCD	608614	4q35.1	Ophthamological
202	Bile acid malabsorption, primary (3)	SLC10A2, NTCP2	601295	13q33	Gastrointestinal
203	Biotinidase deficiency, 253260 (3)	BTD	609019	3p25	Metabolic
204	Bipolar disorder, susceptibility to, 125480 (3)	XBP1, XBP2	194355	22q12	Psychiatric
205	Birt-Hogg-Dube syndrome, 135150 (3)	FLCN, BHD	607273	17p11.2	Dermatological
207	Bladder cancer, 109800 (3)	FGFR3, ACH	134934	4p16.3	Cancer
207	Bladder cancer, 109800 (3)	KRAS2, RASK2	190070	12p12.1	Cancer
207	Bladder cancer, 109800 (3)	RB1	180200	13q14.1-q14.2	Cancer
207	Bladder cancer, somatic, 109800 (3)	HRAS	190020	11p15.5	Cancer
208	Blau syndrome, 186580 (3)	CARD15, NOD2, IBD1, CD, ACUG, PSORAS1	605956	16q12	multiple
209	Bleeding disorder due to defective thromboxane A2 receptor (3)	TBXA2R	188070	19p13.3	Hematological
209	Bleeding due to platelet ADP receptor defect, 600515 (3)	P2RX1, P2X1	600845	17p13.3	Hematological
210	Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 (3)	FOXL2, BPES, BPES1, PFRK, POF3	605597	3q23	multiple
210	Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 (3)	FOXL2, BPES, BPES1, PFRK, POF3	605597	3q23	multiple
211	Blepharospasm, primary benign, 606798 (3)	DRD5, DRD1B, DRD1L2	126453	4p16.1-p15.3	Ophthamological

212	Blood group, ABO system (3)	ABO	110300	9q34	Hematological
212	Blood group, Auberger system (3)	LU, AU, BCAM	111200	19q13.2	Hematological
212	Blood group, Colton, 110450 (3)	AQP1, CHIP28, CO	107776	7p14	Hematological
212	Blood group Cromer (3)	DAF	125240	1q32	Hematological
212	Blood group, Diego, 110500 (3)	SLC4A1, AE1, EPB3	109270	17q21-q22	Hematological
212	Blood group, Dombrock (3)	ART4, DO	110600	12p13-p12	Hematological
212	Blood group, Gerbich (3)	GYPC, GE, GPC	110750	2q14-q21	Hematological
212	Blood group GIL, 607457 (3)	AQP3	600170	9p13	Hematological
212	Blood group, Ii, 110800 (3)	GCNT2	600429	6p24-p23	Hematological
212	Blood group, Indian system (3)	CD44, MDU2, MDU3, MIC4	107269	11pter-p13	Hematological
212	Blood group, Kell (3)	KEL	110900	7q33	Hematological
212	Blood group, Kidd (3)	SLC14A1, JK, UTE, UT1	111000	18q11-q12	Hematological
212	Blood group, Knops system, 607486 (3)	CR1, C3BR	120620	1q32	Hematological
212	Blood group, Landsteiner-Wiener (3)	LW	111250	19p13.3	Hematological
212	Blood group, Lewis (3)	FUT3, LE	111100	19p13.3	Hematological
212	Blood group, Lutheran system (3)	LU, AU, BCAM	111200	19q13.2	Hematological
212	Blood group, MN (3)	GYPA, MN, GPA	111300	4q28.2-q31.1	Hematological
212	Blood group, OK, 111380 (3)	BSG	109480	19p13.3	Hematological
212	Blood group, P system, 111400 (3)	A4GALT, PK	607922	22q13.2	Hematological
212	Blood group, P system, 111400 (3)	B3GALT3, GLCT3, P	603094	3q25	Hematological
212	Blood group, Rhesus (3)	RHCE	111700	1p36.2-p34	Hematological
212	Blood group, Ss (3)	GYPB, SS, MNS	111740	4q28-q31	Hematological
212	Blood group, Waldner, 112010 (3)	SLC4A1, AE1, EPB3	109270	17q21-q22	Hematological
212	Blood group, Wright, 112050 (3)	SLC4A1, AE1, EPB3	109270	17q21-q22	Hematological
212	Blood group, XG system (3)	XG	314700	Xpter-p22.32	Hematological
212	Blood group, Yt system, 112100 (3)	ACHE, YT	100740	7q22	Hematological
213	Bloom syndrome, 210900 (3)	RECQL3, RECQ2, BLM, BS	604610	15q26.1	Cancer
214	Blue-cone monochromacy, 303700 (3)	OPN1LW, RCP, CBP, CBBM	303900	Xq28	Ophthamological
214	Blue-cone monochromacy, 303700 (3)	OPN1MW, GCP, CBD, CBBM	303800	Xq28	Ophthamological
216	Bombay phenotype (3)	FUT1, H, HH	211100	19q13.3	Hematological
216	Bombay phenotype (3)	FUT2, SE	182100	19q13.3	Hematological
217	Bone mineral density variability 1, 601884 (3)	LRP5, BMND1, LRP7, LR3, OPPG, VBCH2	603506	11q13.4	Bone
218	Borjeson-Forssman-Lehmann syndrome, 301900 (3)	PHF6, BFLS	300414	Xq26.3	multiple
220	Bosley-Salih-Alorainy syndrome, 601536 (3)	HOXA1, HOX1F, BSAS	142955	7p15.3	Neurological
221	Bothnia retinal dystrophy, 607475 (3)	RLBP1	180090	15q26	Ophthamological
224	Brachydactyly, type A1, 112500 (3)	IHH, BDA1	600726	2q33-q35	Skeletal
224	Brachydactyly, type A2, 112600 (3)	BMPR1B, ALK6	603248	4q23-q24	Skeletal
224	Brachydactyly, type B1, 113000 (3)	ROR2, BDB1, BDB, NTRKR2	602337	9q22	Skeletal
224	Brachydactyly, type C, 113100 (3)	GDF5, CDMP1	601146	20q11.2	Skeletal
224	Brachydactyly, type D, 113200 (3)	HOXD13, HOX4I, SPD	142989	2q31-q32	Skeletal
224	Brachydactyly, type E, 113300 (3)	HOXD13, HOX4I, SPD	142989	2q31-q32	Skeletal
225	Bradyopsia, 608415 (3)	R9AP, RGS9, PERRS	607814	19q13.11	Ophthamological
225	Bradyopsia, 608415 (3)	RGS9, PERRS	604067	17q23-q24	Ophthamological
226	Branchiootic syndrome (3)	EYA1, BOR	601653	8q13.3	Neurological
226	Branchiootorenal syndrome, 113650 (3)	EYA1, BOR	601653	8q13.3	Neurological
226	Branchiootorenal syndrome with cataract, 113650 (3)	EYA1, BOR	601653	8q13.3	Neurological
228	Breast and colorectal cancer, susceptibility to (3)	CHEK2, RAD53, CHK2, CDS1, LFS2	604373	22q12.1	Cancer
228	Breast cancer, 114480 (3)	PIK3CA	171834	3q26.3	Cancer
228	Breast cancer, 114480 (3)	PPM1D, WIP1	605100	17q22-q23	Cancer

228	Breast cancer, 114480 (3)	SLC22A1L, BWSCR1A, IMPT1	602631	11p15.5	Cancer
228	Breast cancer, 114480 (3)	TP53, P53, LFS1	191170	17p13.1	Cancer
228	Breast cancer-1 (3)	BRCA1, PSCP	113705	17q21	Cancer
228	Breast cancer 2, early onset (3)	BRCA2, FANCD1	600185	13q12.3	Cancer
228	Breast cancer (3)	TSG101	601387	11p15.2-p15.1	Cancer
228	Breast cancer, early-onset, 114480 (3)	BRIP1, BACH1, FANCJ	605882	17q22	Cancer
228	Breast cancer, invasive intraductal (3)	RAD54L, HR54, HRAD54	603615	1p32	Cancer
228	Breast cancer, lobular (3)	CDH1, UVO	192090	16q22.1	Cancer
228	Breast cancer, male, susceptibility to, 114480 (3)	BRCA2, FANCD1	600185	13q12.3	Cancer
228	Breast cancer, male, with Reifenstein syndrome (3)	AR, DHTR, TFM, SBMA, KD, SMAX1	313700	Xq11-q12	Cancer
228	Breast cancer, somatic, 114480 (3)	KRAS2, RASK2	190070	12p12.1	Cancer
228	Breast cancer, somatic, 114480 (3)	RB1CC1, CC1, KIAA0203	606837	8q11	Cancer
228	Breast cancer, sporadic (3)	PHB	176705	17q21	Cancer
228	Breast cancer, susceptibility to, 114480 (3)	ATM, ATA, AT1	607585	11q22.3	Cancer
228	Breast cancer, susceptibility to, 114480 (3)	BARD1	601593	2q34-q35	Cancer
228	Breast cancer, susceptibility to, 114480 (3)	CHEK2, RAD53, CHK2, CDS1, LFS2	604373	22q12.1	Cancer
228	Breast cancer, susceptibility to, 114480 (3)	RAD51A, RECA	179617	15q15.1	Cancer
228	Breast cancer, susceptibility to (3)	XRCC3	600675	14q32.3	Cancer
228	Breast-ovarian cancer (3)	BRCA1, PSCP	113705	17q21	Cancer
231	Brody myopathy, 601003 (3)	ATP2A1, SERCA1	108730	16p12	Muscular
233	Bruck syndrome 2, 609220 (3)	PLOD2	601865	3q23-q24	Unclassified
234	Brugada syndrome, 601144 (3)	SCN5A, LQT3, IVF, HB1, SSS1	600163	3p21	Cardiovascular
235	Brunner syndrome (3)	MAOA	309850	Xp11.23	Unclassified
237	Burkitt lymphoma, 113970 (3)	MYC	190080	8q24.12-q24.13	Cancer
238	Buschke-Ollendorff syndrome, 166700 (3)	LEMD3, MAN1	607844	12q14	multiple
239	Butterfly dystrophy, retinal, 169150 (3)	RDS, RP7, PRPH2, PRPH, AVMD, AOFMD	179605	6p21.1-cen	Ophthamological
240	C1q deficiency, type A (3)	C1QA	120550	1p36.3-p34.1	Immunological
240	C1q deficiency, type B (3)	C1QB	120570	1p36.3-p34.1	Immunological
240	C1q deficiency, type C (3)	C1QG	120575	1p36.3-p34.1	Immunological
240	C1s deficiency, isolated (3)	C1S	120580	12p13	Immunological
240	C2 deficiency (3)	C2	217000	6p21.3	Immunological
240	C3b inactivator deficiency (3)	IF	217030	4q25	Immunological
240	C3 deficiency (3)	C3	120700	19p13.3-p13.2	Immunological
240	C4 deficiency (3)	C4A, C4S	120810	6p21.3	Immunological
240	C4 deficiency (3)	C4B, C4F	120820	6p21.3	Immunological
240	C6 deficiency (3)	C6	217050	5p13	Immunological
240	C7 deficiency (3)	C7	217070	5p13	Immunological
240	C8 deficiency, type II (3)	C8B	120960	1p32	Immunological
240	C9 deficiency (3)	C9	120940	5p13	Immunological
240	C9 deficiency with dermatomyositis (3)	C9	120940	5p13	Immunological
252	Cafe-au-lait spots, multiple, with leukemia, 114030 (3)	MSH2, COCA1, FCC1, HNPCC1	609309	2p22-p21	Cancer
252	Cafe-au-lait spots with glioma or leukemia, 114030 (3)	MLH1, COCA2, HNPCC2	120436	3p21.3	Cancer
253	Caffey disease, 114000 (3)	COL1A1	120150	17q21.31-q22	Connective tissue
254	Calcinosis, tumoral, 211900 (3)	FGF23, ADHR, HPDR2, PHPTC	605380	12p13.3	Bone
254	Calcinosis, tumoral, 211900 (3)	GALNT3	601756	2q24-q31	Bone
255	Campomelic dysplasia, 114290 (3)	SOX9, CMD1, SRA1	608160	17q24.3-q25.1	Skeletal
255	Campomelic dysplasia with autosomal sex reversal, 114290 (3)	SOX9, CMD1, SRA1	608160	17q24.3-q25.1	Skeletal
256	Camptodactyly-arthropathy-coxa vara-pericarditis syndrome, 208250 (3)	PRG4, CACP, MSF, SZP, HAPO	604283	1q24-q25	Skeletal
257	Camurati-Engelmann disease, 131300 (3)	TGFB1, DPD1, CED	190180	19q13.1	Skeletal

258	Canavan disease, 271900 (3)	ASPA	608034	17pter-p13	Metabolic
259	Cancer progression/metastasis (3)	FGFR4	134935	5q35.1-qter	Cancer
259	Cancer susceptibility (3)	MSH6, GTBP, HNPCC5	600678	2p16	Cancer
261	Capillary malformation-arteriovenous malformation, 608354 (3)	RASA1, GAP, CMAVM, PKWS	139150	5q13.3	Cardiovascular
262	Carbamoylphosphate synthetase I deficiency, 237300 (3)	CPS1	608307	2q35	Metabolic
263	Carbohydrate-deficient glycoprotein syndrome, type I, 212065 (3)	PMM2, CDG1	601785	16p13.3-p13.2	Metabolic
263	Carbohydrate-deficient glycoprotein syndrome, type Ib, 602579 (3)	MPI, PMI1	154550	15q22-qter	Metabolic
263	Carbohydrate-deficient glycoprotein syndrome, type II, 212066 (3)	MGAT2, CDGS2	602616	14q21	Metabolic
264	Carboxypeptidase N deficiency, 212070 (3)	CPN1, SCPN, CPN	603103	10q24.2	Hematological
2265	Carcinoid tumor of lung (3)	MEN1	131100	11q13	Cancer
265	Carcinoid tumors, intestinal, 114900 (3)	SDHD, PGL1	602690	11q23	Cancer
267	Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, 604377 (3)	SCO2	604272	22q13	Cardiovascular
268	Cardiomyopathy, Familial hypertrophic, 8, 608751 (3)	MYL3, CMH8	160790	3p	Cardiovascular
268	Cardiomyopathy, dilated, 115200 (3)	ACTC	102540	15q14	Cardiovascular
268	Cardiomyopathy, dilated, 115200 (3)	MYH7, CMH1, MPD1	160760	14q12	Cardiovascular
268	Cardiomyopathy, dilated, 1A, 115200 (3)	LMNA, LMN1, EMD2, FPLD, CMD1A, HGPS, LGMD1B	150330	1q21.2	Cardiovascular
268	Cardiomyopathy, dilated, 1D, 601494 (3)	TNNT2, CMH2, CMD1D	191045	1q32	Cardiovascular
268	Cardiomyopathy, dilated, 1G, 604145 (3), Tibial muscular dystrophy, tardive, 600334 (3)	TTN, CMD1G, TMD, LGMD2J	188840	2q24.3	Cardiovascular
268	Cardiomyopathy, dilated, 1I, 604765 (3)	DES, CMD1I	125660	2q35	Cardiovascular
268	Cardiomyopathy, dilated, 1J, 605362 (3)	EYA4, DFNA10, CMD1J	603550	6q23	Cardiovascular
268	Cardiomyopathy, dilated, 1L, 606685 (3)	SGCD, SGD, LGMD2F, CMD1L	601411	5q33	Cardiovascular
268	Cardiomyopathy, dilated, 1M, 607482 (3)	CSRP3, CRP3, CLP, CMD1M	600824	11p15.1	Cardiovascular
268	Cardiomyopathy, dilated, 1N, 607487 (3)	TCAP, LGMD2G, CMD1N	604488	17q12	Cardiovascular
268	Cardiomyopathy, dilated, with ventricular tachycardia, 608569 (3)	ABCC9, SUR2	601439	12p12.1	Cardiovascular
268	Cardiomyopathy, dilated, X-linked, 302045 (3)	DMD, BMD	300377	Xp21.2	Cardiovascular
268	Cardiomyopathy, familial hypertrophic, 10, 608758 (3)	MYL2, CMH10	160781	12q23-q24.3	Cardiovascular
268	Cardiomyopathy, familial hypertrophic, 1, 192600 (3)	MYH7, CMH1, MPD1	160760	14q12	Cardiovascular
268	Cardiomyopathy, familial hypertrophic, 192600 (3)	ACTC	102540	15q14	Cardiovascular
268	Cardiomyopathy, familial hypertrophic, 192600 (3)	CAV3, LGMD1C	601253	3p25	Cardiovascular
268	Cardiomyopathy, familial hypertrophic, 192600 (3)	MYH6, ASD3, MYHCA	160710	14q12	Cardiovascular
268	Cardiomyopathy, familial hypertrophic, 192600 (3) ()	TNNC1	191040	3p21.3-p14.3	Cardiovascular
268	Cardiomyopathy, familial hypertrophic, 2, 115195 (3)	TNNT2, CMH2, CMD1D	191045	1q32	Cardiovascular
268	Cardiomyopathy, familial hypertrophic, 3, 115196 (3)	TPM1, CMH3	191010	15q22.1	Cardiovascular
268	Cardiomyopathy, familial hypertrophic (3)	TNNI3	191044	19q13.4	Cardiovascular
268	Cardiomyopathy, familial hypertrophic, 4, 115197 (3)	MYBPC3, CMH4	600958	11p11.2	Cardiovascular
268	Cardiomyopathy, familial hypertrophic, 9 (3)	TTN, CMD1G, TMD, LGMD2J	188840	2q24.3	Cardiovascular
268	Cardiomyopathy, familial restrictive, 115210 (3)	TNNI3	191044	19q13.4	Cardiovascular
268	Cardiomyopathy, hypertrophic, early-onset fatal (3)	COX15	603646	10q24	Cardiovascular
268	Cardiomyopathy, hypertrophic, mid-left ventricular chamber type, 608758 (3)	MYL2, CMH10	160781	12q23-q24.3	Cardiovascular
268	Cardiomyopathy, hypertrophic, midventricular, digenic, 192600 (3)	MYLK2, MLCK	606566	20q13.3	Cardiovascular
268	Cardiomyopathy, hypertrophic, with WPW, 600858 (3)	PRKAG2, WPWS	602743	7q36	Cardiovascular
268	Cardiomyopathy, idiopathic dilated, 115200 (3)	PLN, PLB	172405	6q22.1	Cardiovascular
268	Cardiomyopathy, X-linked dilated, 300069 (3)	TAZ, EFE2, BTHS, CMD3A, LVNCX	300394	Xq28	Cardiovascular
269	Carney complex, type 1, 160980 (3)	PRKAR1A, TSE1, CNC1, CAR	188830	17q23-q24	multiple
269	Carney complex variant, 608837 (3)	MYH8	160741	17p13.1	multiple
271	Carnitine-acylcarnitine translocase deficiency (3)	SLC25A20, CACT, CAC	212138	3p21.31	Metabolic
272	Carnitine deficiency, systemic primary, 212140 (3)	SLC22A5, OCTN2, CDSP, SCD	603377	5q31.1	Metabolic

275	Carpal tunnel syndrome, familial (3)	TTR, PALB	176300	18q11.2-q12.1	Unclassified
276	Cartilage-hair hypoplasia, 250250 (3)	RMRP, RMRPR, CHH	157660	9p21-p12	Skeletal
277	Cataract, autosomal dominant nuclear (3)	CRYAA, CRYA1	123580	21q22.3	Ophthamological
277	Cataract, cerulean, type 2, 601547 (3)	CRYBB2, CRYB2	123620	22q11.2-q12.2	Ophthamological
277	Cataract, congenital (3)	PITX3	602669	10q25	Ophthamological
277	Cataract, congenital, 604219 (3)	BFSP2, CP49, CP47	603212	3q21-q25	Ophthamological
277	Cataract, congenital progressive, autosomal recessive (3)	CRYAA, CRYA1	123580	21q22.3	Ophthamological
277	Cataract, congenital, with late-onset corneal dystrophy (3)	PAX6, AN2, MGDA	607108	11p13	Ophthamological
277	Cataract, congenital zonular, with sutural opacities, 600881 (3)	CRYBA1, CRYB1	123610	17q11.1-q12	Ophthamological
277	Cataract, Coppock-like, 604307 (3)	CRYGC, CRYG3, CCL	123680	2q33-q35	Ophthamological
277	Cataract, cortical pulverulent, late-onset (3)	LIM2, MP19	154045	19q13.4	Ophthamological
277	Cataract, crystalline aculeiform, 115700 (3)	CRYGD, CRYG4	123690	2q33-q35	Ophthamological
277	Cataract, juvenile-onset, 604219 (3)	BFSP2, CP49, CP47	603212	3q21-q25	Ophthamological
277	Cataract, lamellar, 116800 (3)	HSF4, CTM	602438	16q21-q22.1	Ophthamological
277	Cataract, Marner type, 116800 (3)	HSF4, CTM	602438	16q21-q22.1	Ophthamological
277	Cataract, polymorphic and lamellar, 604219 (3)	MIP, AQP0	154050	12q13	Ophthamological
277	Cataract, posterior polar 2 (3)	CRYAB, CRYA2, CTPP2	123590	11q22.3-q23.1	Ophthamological
277	Cataract, pulverulent (3)	CRYBB1	600929	22q11.2-q12.1	Ophthamological
277	Cataracts, punctate, progressive juvenile-onset (3)	CRYGD, CRYG4	123690	2q33-q35	Ophthamological
277	Cataract, sutural, with punctate and cerulean opacities, 607133 (3)	CRYBB2, CRYB2	123620	22q11.2-q12.2	Ophthamological
277	Cataract, variable zonular pulverulent (3)	CRYGC, CRYG3, CCL	123680	2q33-q35	Ophthamological
277	Cataract, zonular central nuclear, autosomal dominant (3)	CRYAA, CRYA1	123580	21q22.3	Ophthamological
277	Cataract, zonular pulverulent-1, 116200 (3)	GJA8, CX50, CAE1	600897	1q21.1	Ophthamological
277	Cataract, zonular pulverulent-3, 601885 (3)	GJA3, CX46, CZP3, CAE3	121015	13q11	Ophthamological
279	Cavernous malformations of CNS and retina, 116860 (3)	CCM1, CAM, KRIT1	604214	7q11.2-q21	Cardiovascular
283	CD59 deficiency (3)	CD59, MIC11	107271	11p13	Immunological
284	CD8 deficiency, familial, 608957 (3)	CD8A	186910	2p12	Immunological
2287	Central core disease, 117000 (3)	RYR1, MHS, CCO	180901	19q13.1	Muscular
2287	Central core disease, one form (3) ()	MYH7, CMH1, MPD1	160760	14q12	Muscular
287	Central hypoventilation syndrome, 209880 (3)	GDNF	600837	5p13.1-p12	Respiratory
287	Central hypoventilation syndrome, congenital, 209880 (3)	BDNF	113505	11p13	Respiratory
287	Central hypoventilation syndrome, congenital, 209880 (3)	EDN3	131242	20q13.2-q13.3	Respiratory
287	Central hypoventilation syndrome, congenital, 209880 (3)	PMX2B, NBPHOX, PHOX2B	603851	4p12	Respiratory
287	Central hypoventilation syndrome, congenital, 209880 (3)	RET, MEN2A	164761	10q11.2	Respiratory
289	Cerebellar ataxia, 604290 (3)	CP	117700	3q23-q24	Neurological
289	Cerebellar ataxia, pure (3)	CACNA1A, CACNL1A4, SCA6	601011	19p13	Neurological
4289	Cerebellar hypoplasia, VLDLR-associated, 224050 (3)	VLDLR, VLDLRCH	192977	9p24	Neurological
291	Cerebral amyloid angiopathy, 105150 (3)	ABCA1, ABC1, HDLDT1, TGD	600046	9q22-q31	Neurological
291	Cerebral amyloid angiopathy, 105150 (3)	CST3	604312	20p11.2	Neurological
2291	Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy, 125310 (3)	NOTCH3, CADASIL, CASIL	600276	19p13.2-p13.1	Cardiovascular
4291	Cerebral cavernous malformations-1, 116860 (3)	CCM1, CAM, KRIT1	604214	7q11.2-q21	Neurological
4291	Cerebral cavernous malformations-2, 603284 (3)	C7orf22, CCM2, MGC4067	607929	7p13	Neurological
4291	Cerebral cavernous malformations 3, 603285 (3)	PDCD10, TFAR15, CCM3	609118	3q26.1	Neurological
6291	Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528 (3)	SNAP29, CEDNIK	604202	22q11.2	Neurological
292	Cerebrooculofacioskeletal syndrome, 214150 (3)	ERCC2, EM9	126340	19q13.2-q13.3	multiple
292	Cerebrooculofacioskeletal syndrome, 214150 (3)	ERCC5, XPG	133530	13q33	multiple
292	Cerebrooculofacioskeletal syndrome 214150 (3)	ERCC6, CKN2, COFS, CSB	609413	10q11	multiple
293	Cerebrotendinous xanthomatosis, 213700 (3)	CYP27A1, CYP27, CTX	606530	2q33-qter	Metabolic

294	Cerebrovascular disease, occlusive (3)	SERPINA3, AACT, ACT	107280	14q32.1	Unclassified
295	Ceroid lipofuscinosis, neuronal-1, infantile, 256730 (3)	PPT1, CLN1	600722	1p32	Neurological
296	Ceroid-lipofuscinosis, neuronal 2, classic late infantile, 204500 (3)	CLN2	607998	11p15.5	Neurological
296	Ceroid-lipofuscinosis, neuronal-3, juvenile, 204200 (3)	CLN3, BTS	607042	16p12.1	Neurological
296	Ceroid-lipofuscinosis, neuronal-5, variant late infantile, 256731 (3)	CLN5	608102	13q21.1-q32	Neurological
296	Ceroid-lipofuscinosis, neuronal-6, variant late infantile, 601780 (3)	CLN6	606725	15q21-q23	Neurological
296	Ceroid lipofuscinosis, neuronal 8, 600143 (3)	CLN8, EPMR	607837	8pter-p22	Neurological
296	Ceroid lipofuscinosis, neuronal, variant juvenile type, with granular osmiophilic deposits (3)	PPT1, CLN1	600722	1p32	Neurological
298	Cervical cancer, somatic, 603956 (3)	FGFR3, ACH	134934	4p16.3	Cancer
299	CETP deficiency, 607322 (3)	CETP	118470	16q21	Metabolic
300	Chanarin-Dorfman syndrome, 275630 (3)	ABHD5, CGI58, IECN2, NCIE2	604780	3p21	Metabolic
301	Charcot-Marie-Tooth disease, axonal, type 2F, 606595 (3)	HSPB1, HSP27, CMT2F	602195	7q11.23	Neurological
301	Charcot-Marie-Tooth disease, dominant intermediate 3, 607791 (3)	MPZ, CMT1B, CMTDI3, CHM, DSS	159440	1q22	Neurological
301	Charcot-Marie-Tooth disease, dominant intermediate B, 606482 (3)	DNM2	602378	19p13.2	Neurological
301	Charcot-Marie-Tooth disease, foot deformity of (3)	HOXD10, HOX4D	142984	2q31-q32	Neurological
301	Charcot-Marie-Tooth disease, mixed axonal and demyelinating type, 214400 (3)	GDAP1, CMT4A, CMT2K, CMT2G	606598	8q13-q21.1	Neurological
301	Charcot-Marie-Tooth disease, type 1A, 118220 (3)	PMP22, CMT1A, CMT1E, DSS	601097	17p11.2	Neurological
301	Charcot-Marie-Tooth disease, type 1B, 118200 (3)	MPZ, CMT1B, CMTDI3, CHM, DSS	159440	1q22	Neurological
301	Charcot-Marie-Tooth disease, type 1C, 601098 (3)	LITAF, CMT1C	603795	16p13.3-p12	Neurological
301	Charcot-Marie-Tooth disease, type 1D, 607678 (3)	EGR2, KROX20	129010	10q21.1-q22.1	Neurological
301	Charcot-Marie-Tooth disease, type 1E, 118300 (3)	PMP22, CMT1A, CMT1E, DSS	601097	17p11.2	Neurological
301	Charcot-Marie-Tooth disease, type 1F, 607734 (3)	NEFL, CMT2E, CMT1F	162280	8p21	Neurological
301	Charcot-Marie-Tooth disease, type 2A1, 118210 (3)	KIF1B, CMT2A, CMT2A1	605995	1p36.2	Neurological
301	Charcot-Marie-Tooth disease, type 2A2, 609260 (3)	MFN2, KIAA0214, CMT2A2	608507	1p36.2	Neurological
301	Charcot-Marie-Tooth disease, type 2B, 600882 (3)	RAB7, CMT2B, PSN	602298	3q21	Neurological
301	Charcot-Marie-Tooth disease, type 2D, 601472 (3)	GARS, SMAD1, CMT2D	600287	7p15	Neurological
301	Charcot-Marie-Tooth disease, type 2E, 607684 (3)	NEFL, CMT2E, CMT1F	162280	8p21	Neurological
301	Charcot-Marie-Tooth disease, type 2G, 607706 (3)	GDAP1, CMT4A, CMT2K, CMT2G	606598	8q13-q21.1	Neurological
301	Charcot-Marie-Tooth disease, type 2I, 607677 (3)	MPZ, CMT1B, CMTDI3, CHM, DSS	159440	1q22	Neurological
301	Charcot-Marie-Tooth disease, type 2J, 607736 (3)	MPZ, CMT1B, CMTDI3, CHM, DSS	159440	1q22	Neurological
301	Charcot-Marie-Tooth disease, type 2K, 607831 (3)	GDAP1, CMT4A, CMT2K, CMT2G	606598	8q13-q21.1	Neurological
301	Charcot-Marie-Tooth disease, type 4A, 214400 (3)	GDAP1, CMT4A, CMT2K, CMT2G	606598	8q13-q21.1	Neurological
301	Charcot-Marie-Tooth disease, type 4B1, 601382 (3)	MTMR2, CMT4B1	603557	11q22	Neurological
301	Charcot-Marie-Tooth disease, type 4B2, 604563 (3)	SBF2, MTMR13, CMT4B2	607697	11p15	Neurological
301	Charcot-Marie-Tooth disease, type 4B2, with early-onset glaucoma, 607739 (3)	SBF2, MTMR13, CMT4B2	607697	11p15	Neurological
301	Charcot-Marie-Tooth disease, type 4C, 601596 (3)	KIAA1985	608206	5q32	Neurological
301	Charcot-Marie-Tooth disease, type 4D, 601455 (3)	NDRG1, HMSNL, CMT4D	605262	8q24.3	Neurological
301	Charcot-Marie-Tooth neuropathy, X-linked dominant, 1, 302800 (3)	GJB1, CX32, CMTX1	304040	Xq13.1	Neurological
302	CHARGE syndrome, 214800 (3)	CHD7	608892	8q12.1	multiple
303	Char syndrome, 169100 (3)	TFAP2B, CHAR	601601	6p12	multiple
304	Chediak-Higashi syndrome, 214500 (3)	CHS1, LYST	606897	1q42.1-q42.2	multiple
305	Cherubism, 118400 (3)	SH3BP2, CRPM	602104	4p16.3	Unclassified
306	CHILD syndrome, 308050 (3)	NSDHL	300275	Xq28	Developmental
307	Chitotriosidase deficiency (3)	CHIT	600031	1q31-q32	Metabolic
308	Chloride diarrhea, congenital, Finnish type, 214700 (3)	SLC26A3, DRA, CLD	126650	7q22-q31.1	Gastrointestinal
309	Cholelithiasis, 600803 (3)	ABCB4, PGY3, MDR3	171060	7q21.1	Gastrointestinal
310	Cholestasis, benign recurrent intrahepatic, 243300 (3)	ATP8B1, FIC1, BRIC, PFIC1	602397	18q21	Gastrointestinal

310	Cholestasis, familial intrahepatic, of pregnancy, 147480 (3)	ABCB4, PGY3, MDR3	171060	7q21.1	Gastrointestinal
310	Cholestasis, progressive familial intrahepatic 1, 211600 (3)	ATP8B1, FIC1, BRIC, PFIC1	602397	18q21	Gastrointestinal
310	Cholestasis, progressive familial intrahepatic 2, 601847 (3)	ABCB11, BSEP, SPGP, PFIC2	603201	2q24	Gastrointestinal
310	Cholestasis, progressive familial intrahepatic 3, 602347 (3)	ABCB4, PGY3, MDR3	171060	7q21.1	Gastrointestinal
310	Cholestasis, progressive familial intrahepatic 4, 607765 (3)	HSD3B7, PFIC4	607764	16p12-p11.2	Gastrointestinal
313	Cholesteryl ester storage disease (3)	LIPA	278000	10q24-q25	Metabolic
314	Chondrocalcinosis 2, 118600 (3)	ANKH, HANK, ANK, CMDJ, CCAL2, CPPDD	605145	5p15.2-p14.1	Connective tissue disorder
2315	Chondrodysplasia, Grebe type, 200700 (3)	GDF5, CDMP1	601146	20q11.2	Skeletal
315	Chondrodysplasia punctata, rhizomelic, type 2, 222765 (3)	GNPAT, DHAPAT	602744	1q42	Connective tissue
315	Chondrodysplasia punctata, X-linked dominant, 302960 (3)	EBP, CDPX2, CPXD, CPX	300205	Xp11.23-p11.22	Connective tissue
315	Chondrodysplasia punctata, X-linked recessive, 302950 (3)	ARSE, CDPX1, CDPXR	300180	Xp22.3	Connective tissue
316	Chondrosarcoma, 215300 (3)	EXT1	608177	8q24.11-q24.13	Cancer
316	Chondrosarcoma, extraskeletal myxoid (3)	CSMF	600542	9q22	Cancer
316	Chondrosarcoma, extraskeletal myxoid (3)	EWSR1, EWS	133450	22q12	Cancer
318	Chorea, hereditary benign, 118700 (3)	TITF1, NKX2A, TTF1	600635	14q13	Neurological
319	Choreoacanthocytosis, 200150 (3)	VPS13A, CHAC	605978	9q21	Neurological
320	Choreoathetosis, hypothyroidism, and respiratory distress (3)	TITF1, NKX2A, TTF1	600635	14q13	multiple
323	Choroideremia, 303100 (3)	CHM, TCD	300390	Xq21.2	Ophthamological
326	Chromosome 22q13.3 deletion syndrome, 606232 (3)	PSAP2, PROSAP2, KIAA1650	606230	22q13.3	multiple
327	Chronic granulomatous disease, autosomal, due to deficiency of CYBA, 233690 (3)	CYBA	608508	16q24	Immunological
327	Chronic granulomatous disease due to deficiency of NCF-1, 233700 (3)	NCF1	608512	7q11.23	Immunological
327	Chronic granulomatous disease due to deficiency of NCF-2, 233710 (3)	NCF2	608515	1q25	Immunological
327	Chronic granulomatous disease, X-linked, 306400 (3)	CYBB, CGD	300481	Xp21.1	Immunological
2327	Chronic infections, due to opsonin defect (3)	MBL2, MBL, MBP1	154545	10q11.2-q21	Immunological
328	Chudley-Lowry syndrome, 309490 (3)	ATRX, XH2, XNP, MRXS3, SHS	300032	Xq13	multiple
329	Chylomicronemia syndrome, familial (3)	LPL, LIPD	238600	8p22	Metabolic
330	Chylomicron retention disease, 246700 (3)	SARA2, SAR1B, CMRD	607690	5q31.1	Gastrointestinal
330	Chylomicron retention disease with Marinesco-Sjogren syndrome, 607692 (3)	SARA2, SAR1B, CMRD	607690	5q31.1	Gastrointestinal
331	Ciliary dyskinesia, primary, 1, 242650 (3)	DNAI1, CILD1, ICS, PCD	604366	9p21-p13	Respiratory
331	Ciliary dyskinesia, primary, 3 608644 (3)	DNAH5, HL1, PCD, CILD3	603335	5p15-p14	Respiratory
332	CINCA syndrome, 607115 (3)	CIAS1, C1orf7, FCU, FCAS	606416	1q44	multiple
334	Cirrhosis, cryptogenic (3)	KRT18	148070	12q13	Gastrointestinal
334	Cirrhosis, cryptogenic (3)	KRT8	148060	12q13	Gastrointestinal
334	Cirrhosis, noncryptogenic, susceptibility to, 215600 (3)	KRT18	148070	12q13	Gastrointestinal
334	Cirrhosis, noncryptogenic, susceptibility to, 215600 (3)	KRT8	148060	12q13	Gastrointestinal
334	Cirrhosis, North American Indian childhood type, 604901 (3)	CIRH1A, NAIC, TEX292, KIAA1988	607456	16q22	Gastrointestinal
335	Citrullinemia, 215700 (3)	ASS	603470	9q34	Metabolic
335	Citrullinemia, adult-onset type II, 603471 (3)	SLC25A13, CTLN2	603859	7q21.3	Metabolic
335	Citrullinemia, type II, neonatal-onset, 605814 (3)	SLC25A13, CTLN2	603859	7q21.3	Metabolic
336	Cleft lip/palate ectodermal dysplasia syndrome, 225000 (3)	HVEC, PVRL1, PVRR1, PRR1	600644	11q23-q24	Developmental
336	Cleft lip/palate, nonsyndromic, 608874 (3)	MSX1, HOX7, HYD1, OFC5	142983	4p16.1	Developmental
336	Cleft palate with ankyloglossia, 303400 (3)	TBX22, CPX	300307	Xq12-q21	Developmental
337	Cleidocranial dysplasia, 119600 (3)	RUNX2, CBFA1, PEBP2A1, AML3	600211	6p21	Skeletal
338	Coats disease, 300216 (3)	NDP, ND	310600	Xp11.4	Ophthamological
339	Cockayne syndrome, type A, 216400 (3)	ERCC8, CKN1, CSA	609412	5q12	multiple
339	Cockayne syndrome, type B, 133540 (3)	ERCC6, CKN2, COFS, CSB	609413	10q11	multiple
340	Codeine sensitivity (3)	CYP2D@, CYP2D, P450C2D	124030	22q13.1	Metabolic
341	Coffin-Lowry syndrome, 303600 (3)	RPS6KA3, RSK2, MRX19	300075	Xp22.2-p22.1	multiple

342	Cohen syndrome, 216550 (3)	COH1	607817	8q22-q23	multiple
343	Colchicine resistance (3)	ABCB1, PGY1, MDR1	171050	7q21.1	Unclassified
344	Cold-induced autoinflammatory syndrome, familial, 120100 (3)	CIAS1, C1orf7, FCU, FCAS	606416	1q44	Immunological
2344	Cold-induced sweating syndrome, 272430 (3)	CRLF1, CISS	604237	19p12	multiple
345	Coloboma, ocular, 120200 (3)	PAX6, AN2, MGDA	607108	11p13	Ophthamological
345	Coloboma, ocular, 120200 (3)	SHH, HPE3, HLP3, SMMCI	600725	7q36	Ophthamological
346	Colon adenocarcinoma (3)	RAD54B	604289	8q21.3-q22	Cancer
346	Colon adenocarcinoma (3)	RAD54L, HR54, HRAD54	603615	1p32	Cancer
346	Colon cancer (3)	BCL10	603517	1p22	Cancer
346	Colon cancer (3)	PTPN12, PTPG1	600079	7q11.23	Cancer
346	Colon cancer (3)	TGFBR2, HNPCC6	190182	3p22	Cancer
346	Colon cancer, advanced (3)	SRC, ASV, SRC1	190090	20q12-q13	Cancer
346	Colon cancer, hereditary nonpolypopsis, type 7 (3)	MLH3, HNPCC7	604395	14q24.3	Cancer
346	Colon cancer, somatic, 114500 (3)	PTPRJ, DEP1	600925	11p11.2	Cancer
346	Colonic adenoma recurrence, reduced risk of, 114500 (3)	ODC1	165640	2p25	Cancer
347	Colonic aganglionosis, total, with small bowel involvement (3)	RET, MEN2A	164761	10q11.2	Gastrointestinal
348	Colorblindness, deutan (3)	OPN1MW, GCP, CBD, CBBM	303800	Xq28	Ophthamological
348	Colorblindness, protan (3)	OPN1LW, RCP, CBP, CBBM	303900	Xq28	Ophthamological
348	Colorblindness, tritan (3)	OPN1SW, BCP, CBT	190900	7q31.3-q32	Ophthamological
346	Colorectal adenomatous polyposis, autosomal recessive, with pilomatricomas, 132600 (3)	MUTYH	604933	1p34.3-p32.1	Cancer
346	Colorectal cancer, 114500 (3)	AXIN2	604025	17q24	Cancer
346	Colorectal cancer, 114500 (3)	BUB1B, BUBR1	602860	15q15	Cancer
346	Colorectal cancer, 114500 (3)	EP300	602700	22q13	Cancer
346	Colorectal cancer, 114500 (3)	PDGFRL, PDGRL, PRLTS	604584	8p22-p21.3	Cancer
346	Colorectal cancer, 114500 (3)	PIK3CA	171834	3q26.3	Cancer
346	Colorectal cancer, 114500 (3)	TP53, P53, LFS1	191170	17p13.1	Cancer
346	Colorectal cancer (3)	APC, GS, FPC	175100	5q21-q22	Cancer
346	Colorectal cancer (3)	BAX	600040	19q13.3-q13.4	Cancer
346	Colorectal cancer (3)	CTNNB1	116806	3p22-p21.3	Cancer
346	Colorectal cancer (3)	DCC	120470	18q21.3	Cancer
346	Colorectal cancer (3)	MCC	159350	5q21	Cancer
346	Colorectal cancer (3)	NRAS	164790	1p13.2	Cancer
346	Colorectal cancer, hereditary nonpolyposis, type 1, 120435 (3)	MSH2, COCA1, FCC1, HNPCC1	609309	2p22-p21	Cancer
346	Colorectal cancer, hereditary nonpolyposis, type 2, 609310 (3)	MLH1, COCA2, HNPCC2	120436	3p21.3	Cancer
346	Colorectal cancer, hereditary nonpolyposis, type 3 (3)	PMS1, PMSL1, HNPCC3	600258	2q31-q33	Cancer
346	Colorectal cancer, hereditary nonpolyposis, type 4 (3)	PMS2, PMSL2, HNPCC4	600259	7p22	Cancer
346	Colorectal cancer, hereditary nonpolyposis, type 5 (3)	MSH6, GTBP, HNPCC5	600678	2p16	Cancer
346	Colorectal cancer, hereditary nonpolyposis, type 6 (3)	TGFBR2, HNPCC6	190182	3p22	Cancer
346	Colorectal cancer, somatic, 109800 (3)	FGFR3, ACH	134934	4p16.3	Cancer
346	Colorectal cancer, somatic, 114500 (3)	FLCN, BHD	607273	17p11.2	Cancer
346	Colorectal cancer, somatic, 114500 (3)	MLH3, HNPCC7	604395	14q24.3	Cancer
346	Colorectal cancer, somatic (3)	BRAF	164757	7q34	Cancer
346	Colorectal cancer, somatic (3)	DLC1	604258	8p22-p21.3	Cancer
346	Colorectal cancer, sporadic, 114500 (3)	PLA2G2A, PLA2B, PLA2L, MOM1	172411	1p35	Cancer
346	Colorectal cancer, susceptibility to (3)	CCND1, PRAD1, BCL1	168461	11q13	Cancer
346	Colorectal cancer with chromosomal instability (3)	BUB1	602452	2q14	Cancer
4350	Combined C6/C7 deficiency (3)	C6	217050	5p13	Immunological
350	Combined factor V and VIII deficiency, 227300 (3)	LMAN1, ERGIC53, F5F8D, MCFD1	601567	18q21.3-q22	Hematological
2350	Combined hyperlipemia, familial (3)	LPL, LIPD	238600	8p22	Metabolic

4350	Combined immunodeficiency, X-linked, moderate, 312863 (3)	IL2RG, SCIDX1, SCIDX, IMD4	308380	Xq13	Immunological
6350	Combined oxidative phosphorylation deficiency, 609060 (3)	GFM1, EFG1, GFM	606639	3q25.1-q26.2	multiple
8350	Combined SAP deficiency (3)	PSAP, SAP1	176801	10q22.1	Metabolic
351	Complex I, mitochondrial respiratory chain, deficiency of, 252010 (3)	NDUFS6	603848	5pter-p15.33	multiple
351	Complex V, mitochondrial respiratory chain, deficiency of, 604273 (3)	ATPAF2, ATP12	608918	17p11.2	multiple
352	Cone dystrophy-1, 304020 (3)	RPGR, RP3, CRD, RP15, COD1	312610	Xp21.1	Ophthamological
352	Cone dystrophy-3, 602093 (3)	GUCA1A, GCAP	600364	6p21.1	Ophthamological
352	Cone-rod dystrophy, 300029 (3)	RPGR, RP3, CRD, RP15, COD1	312610	Xp21.1	Ophthamological
352	Cone-rod dystrophy 3 (3)	ABCA4, ABCR, STGD1, FFM, RP19	601691	1p21-p13	Ophthamological
352	Cone-rod dystrophy (3)	AIPL1, LCA4	604392	17p13.1	Ophthamological
352	Cone-rod dystrophy 6, 601777(3)	GUCY2D, GUC2D, LCA1, CORD6	600179	17p13.1	Ophthamological
352	Cone-rod dystrophy 9, 608194 (3)	RPGRIP1, LCA6, CORD9	605446	14q11	Ophthamological
352	Cone-rod retinal dystrophy-2, 120970 (3)	CRX, CORD2, CRD	602225	19q13.3	Ophthamological
2354	Congenital bilateral absence of vas deferens, 277180 (3)	CFTR, ABCC7, CF, MRP7	602421	7q31.2	Unclassified
4354	Congenital cataracts, facial dysmorphism, and neuropathy, 604168 (3)	CTDP1, FCP1, CCFDN	604927	18q23	Ophthamological
354	Congenital disorder of glycosylation, type Ic, 603147 (3)	ALG6	604566	1p22.3	Metabolic
354	Congenital disorder of glycosylation, type Id, 601110 (3)	ALG3, NOT56L, CDGS4	608750	3q27	Metabolic
354	Congenital disorder of glycosylation, type Ie, 608799 (3)	DPM1, MPDS, CDGIE	603503	20q13.13	Metabolic
354	Congenital disorder of glycosylation, type If, 609180 (3)	MPDU1, SL15, CDGIF	604041	17p13.1-p12	Metabolic
354	Congenital disorder of glycosylation, type Ig, 607143 (3)	ALG12	607144	Chr.22	Metabolic
354	Congenital disorder of glycosylation, type Ih, 608104 (3)	ALG8	608103	11pter-p15.5	Metabolic
354	Congenital disorder of glycosylation, type Ii, 607906 (3)	ALG2, CDGII	607905	9q22	Metabolic
354	Congenital disorder of glycosylation, type II, 608776 (3)	DIBD1, ALG9	606941	11q23	Metabolic
354	Congenital disorder of glycosylation, type IIc, 266265 (3)	SLC35C1, FUCT1	605881	11p11.2	Metabolic
354	Congenital disorder of glycosylation, type IId, 607091 (3)	B4GALT1, GGTB2, GT1, GTB	137060	9p13	Metabolic
354	Congenital disorder of glycosylation, type IIe, 608779 (3)	COG7, CDG2E	606978	16p	Metabolic
354	Congenital disorder of glycosylation, type Ij, 608093 (3)	DPAGT2, DGPT	191350	11q23.3	Metabolic
354	Congenital disorder of glycosylation, type Ik, 608540 (3)	ALG1, HMAT1, HMT1	605907	16p13.3	Metabolic
355	Congestive heart failure, susceptibility to (3)	ADRA2C, ADRA2L2	104250	4p16.1	Cardiovascular
355	Congestive heart failure, susceptibility to (3)	ADRB1, ADRB1R, RHR	109630	10q24-q26	Cardiovascular
356	Conjunctivitis, ligneous, 217090 (3)	PLG	173350	6q26	Ophthamological
357	Conotruncal anomaly face syndrome, 217095 (3)	TBX1, DGS, CTHM, CAFS, TGA, DORV, VCFS, DGCR	602054	22q11.2	Cardiovascular
358	Contractural arachnodactyly, congenital (3)	FBN2, CCA	121050	5q23-q31	Connective tissue
359	Convulsions, familial febrile, 4, 604352 (3)	MASS1, VLGR1, KIAA0686, FEB4, USH2C	602851	5q14	Neurological
360	COPD, rate of decline of lung function in, 606963 (3)	MMP1, CLG	120353	11q22-q23	Respiratory
361	Coproporphyria (3)	CPO	121300	3q12	Metabolic
362	Corneal clouding, autosomal recessive (3)	APOA1	107680	11q23	Ophthamological
362	Corneal dystrophy, Avellino type, 607541 (3)	TGFBI, CSD2, CDGG1, CSD, BIGH3, CDG2	601692	5q31	Ophthamological
362	Corneal dystrophy, gelatinous drop-like, 204870 (3)	TACSTD2, TROP2, M1S1	137290	1p32	Ophthamological
362	Corneal dystrophy, Groenouw type I, 121900 (3)	TGFBI, CSD2, CDGG1, CSD, BIGH3, CDG2	601692	5q31	Ophthamological
362	Corneal dystrophy, hereditary polymorphous posterior, 122000 (3)	VSX1, RINX, PPCD, PPD, KTCN	605020	20p11.2	Ophthamological
362	Corneal dystrophy, hereditary polymorphous posterior, 2, 122000 (3)	COL8A2, FECD, PPCD2	120252	1p34.3-p32.3	Ophthamological
362	Corneal dystrophy, lattice type I, 122200 (3)	TGFBI, CSD2, CDGG1, CSD, BIGH3, CDG2	601692	5q31	Ophthamological
362	Corneal dystrophy, lattice type IIIA, 608471 (3)	TGFBI, CSD2, CDGG1, CSD, BIGH3, CDG2	601692	5q31	Ophthamological

362	Corneal dystrophy, Reis-Bucklers type, 608470 (3)	TGFBI, CSD2, CDGG1, CSD, BIGH3, CDG2	601692	5q31	Ophthamological
362	Corneal dystrophy, Thiel-Behnke type, 602082 (3)	TGFBI, CSD2, CDGG1, CSD, BIGH3, CDG2	601692	5q31	Ophthamological
362	Corneal fleck dystrophy, 121850 (3)	PIP5K3, CFD	609414	2q35	Ophthamological
362	Cornea plana congenita, recessive, 217300 (3)	KERA, CNA2	603288	12q22	Ophthamological
364	Cornelia de Lange syndrome, 122470 (3)	NIPBL, CDLS	608667	5p13.1	Developmental
365	Coronary artery disease, autosomal dominant, 1, 608320 (3)	MEF2A, ADCAD1	600660	15q26	Cardiovascular
365	Coronary artery disease in familial hypercholesterolemia, protection against, 143890 (3)	ABCA1, ABC1, HDLDT1, TGD	600046	9q22-q31	Cardiovascular
365	Coronary artery disease, susceptibility to (3)	KL	604824	13q12	Cardiovascular
365	Coronary artery disease, susceptibility to (3)	PON1, PON, ESA	168820	7q21.3	Cardiovascular
365	Coronary artery disease, susceptibility to (3)	PON2	602447	7q21.3	Cardiovascular
365	Coronary artery spasm, susceptibility to (3)	PON1, PON, ESA	168820	7q21.3	Cardiovascular
365	Coronary heart disease, susceptibility to (3)	MMP3, STMY1	185250	11q23	Cardiovascular
2365	Coronary spasms, susceptibility to (3)	NOS3	163729	7q36	Cardiovascular
366	Corpus callosum, agenesis of, with mental retardation, ocular coloboma and micrognathia, 300472 (3)	IGBP1	300139	Xq13.1-q13.3	Neurological
367	Cortisol resistance (3)	NR3C1, GCR, GRL	138040	5q31	Endocrine
368	Cortisone reductase deficiency, 604931 (3)	GDH	138090	1pter-p36.13	Metabolic
368	Cortisone reductase deficiency, 604931 (3)	HSD11B1, HSD11, HSD11L	600713	1q32-q41	Metabolic
369	Costello syndrome, 218040 (3)	HRAS	190020	11p15.5	multiple
370	Coumarin resistance, 122700 (3)	CYP2A6, CYP2A3, CYP2A, P450C2A	122720	19q13.2	Hematological
372	Cowden disease, 158350 (3)	PTEN, MMAC1	601728	10q23.31	Cancer
372	Cowden-like syndrome, 158350 (3)	BMPR1A, ACVRLK3, ALK3	601299	10q22.3	Cancer
374	CPT deficiency, hepatic, type IA, 255120 (3)	CPT1A	600528	11q13	Metabolic
374	CPT deficiency, hepatic, type II, 600649 (3)	CPT2	600650	1p32	Metabolic
374	CPT II deficiency, lethal neonatal, 608836 (3)	CPT2	600650	1p32	Metabolic
376	Cramps, familial, potassium-aggravated (3)	SCN4A, HYPP, NAC1A	603967	17q23.1-q25.3	Muscular
377	Craniofacial anomalies, empty sella turcica, corneal endothelial changes, and abnormal retinal and auditory bipolar cells (3)	VSX1, RINX, PPCD, PPD, KTCN	605020	20p11.2	multiple
378	Craniofacial-deafness-hand syndrome, 122880 (3)	PAX3, WS1, HUP2, CDHS	606597	2q35	multiple
379	Craniofacial-skeletal-dermatologic dysplasia (3)	FGFR2, BEK, CFD1, JWS	176943	10q26	multiple
380	Craniofrontonasal dysplasia, 304110 (3)	EFNB1, EPLG2, CFNS, CFND	300035	Xq12	Skeletal
382	Craniometaphyseal dysplasia, 123000 (3)	ANKH, HANK, ANK, CMDJ, CCAL2, CPPDD	605145	5p15.2-p14.1	Bone
383	Craniosynostosis, nonspecific (3)	FGFR2, BEK, CFD1, JWS	176943	10q26	Skeletal
383	Craniosynostosis, type 2, 604757 (3)	MSX2, CRS2, HOX8	123101	5q34-q35	Skeletal
384	CRASH syndrome, 303350 (3)	L1CAM, CAML1, HSAS1	308840	Xq28	multiple
2385	Creatine deficiency syndrome, X-linked, 300352 (3)	SLC6A8, CRTR	300036	Xq28	Neurological
385	Creatine phosphokinase, elevated serum, 123320 (3)	CAV3, LGMD1C	601253	3p25	Metabolic
385	Creatine phosphokinase, elevated serum, 123320 (3)	CAV3, LGMD1C	601253	3p25	Metabolic
388	Creutzfeldt-Jakob disease, 123400 (3)	PRNP, PRIP	176640	20pter-p12	Neurological
388	Creutzfeldt-Jakob disease, variant, resistance to, 123400 (3)	HLA-DQB1	604305	6p21.3	Neurological
389	Crigler-Najjar syndrome, type I, 218800 (3)	UGT1A1, UGT1, GNT1	191740	2q37	multiple
389	Crigler-Najjar syndrome, type II, 606785 (3)	UGT1A1, UGT1, GNT1	191740	2q37	multiple
390	Crohn disease, susceptibility to, 266600 (3)	CARD15, NOD2, IBD1, CD, ACUG, PSORAS1	605956	16q12	Gastrointestinal
390	Crohn disease, susceptibility to, 266600 (3)	DLG5, PDLG, KIAA0583	604090	10q23	Gastrointestinal
391	Crouzon syndrome, 123500 (3)	FGFR2, BEK, CFD1, JWS	176943	10q26	Skeletal

391	Crouzon syndrome with acanthosis nigricans (3)	FGFR3, ACH	134934	4p16.3	Skeletal
392	Cryptorchidism, bilateral, 219050 (3)	LGR8, GREAT	606655	13q13.1	Renal
392	Cryptorchidism, idiopathic, 219050 (3)	INSL3	146738	19p13.2	Renal
394	Currarino syndrome, 176450 (3)	HLXB9, HOXHB9, SCRA1	142994	7q36	Skeletal
395	Cutis laxa, AD, 123700 (3)	ELN	130160	7q11.2	Connective tissue
395	Cutis laxa, autosomal dominant, 123700 (3)	FBLN5, ARMD3	604580	14q32.1	Connective tissue
395	Cutis laxa, autosomal recessive, 219100 (3)	FBLN5, ARMD3	604580	14q32.1	Connective tissue
395	Cutis laxa, neonatal (3)	ATP7A, MNK, MK, OHS	300011	Xq12-q13	Connective tissue
396	Cyclic ichthyosis with epidermolytic hyperkeratosis, 607602 (3)	KRT1	139350	12q13	Dermatological
397	Cylindromatosis, familial, 132700 (3)	CYLD1, CDMT, EAC	605018	16q12-q13	Dermatological
398	Cystathioninuria, 219500 (3)	CTH	607657	1p31.1	Metabolic
399	Cystic fibrosis, 219700 (3)	CFTR, ABCC7, CF, MRP7	602421	7q31.2	Respiratory
400	Cystinosis, atypical nephropathic (3)	CTNS	606272	17p13	Renal
400	Cystinosis, late-onset juvenile or adolescent nephropathic, 219900 (3)	CTNS	606272	17p13	Renal
400	Cystinosis, nephropathic, 219800 (3)	CTNS	606272	17p13	Renal
400	Cystinosis, ocular nonnephropathic, 219750 (3)	CTNS	606272	17p13	Renal
401	Cystinuria, 220100 (3)	SLC3A1, ATR1, D2H, NBAT	104614	2p16.3	Renal
401	Cystinuria, type II (3)	SLC7A9, CSNU3	604144	19q13.1	Renal
401	Cystinuria, type III (3)	SLC7A9, CSNU3	604144	19q13.1	Renal
402	D-2-hydroxyglutaric aciduria, 600721 (3)	D2HGD	609186	2p25.3	Metabolic
404	Darier disease, 124200 (3)	ATP2A2, ATP2B, DAR	108740	12q23-q24.1	Dermatological
405	D-bifunctional protein deficiency, 261515 (3)	HSD17B4	601860	5q2	Metabolic
406	Deafness, autosomal dominant 10, 601316 (3)	EYA4, DFNA10, CMD1J	603550	6q23	Ear,Nose,Throat
406	Deafness, autosomal dominant 1, 124900 (3)	DIAPH1, DFNA1, LFHL1	602121	5q31	Ear,Nose,Throat
406	Deafness, autosomal dominant 11, neurosensory, 601317 (3)	MYO7A, USH1B, DFNB2, DFNA11	276903	11q13.5	Ear,Nose,Throat
406	Deafness, autosomal dominant 12, 601842 (3)	TECTA, DFNA8, DFNA12, DFNB21	602574	11q22-q24	Ear,Nose,Throat
406	Deafness, autosomal dominant 13, 601868 (3)	COL11A2, STL3, DFNA13	120290	6p21.3	Ear,Nose,Throat
406	Deafness, autosomal dominant 15, 602459 (3)	POU4F3, BRN3C	602460	5q31	Ear,Nose,Throat
406	Deafness, autosomal dominant 17, 603622 (3)	MYH9, MHA, FTNS, DFNA17	160775	22q11.2	Ear,Nose,Throat
406	Deafness, autosomal dominant 20/26, 604717 (3)	ACTG1, DFNA20, DFNA26	102560	17q25.3	Ear,Nose,Throat
406	Deafness, autosomal dominant 22, 606346 (3)	MYO6, DFNA22, DFNB37	600970	6q13	Ear,Nose,Throat
406	Deafness, autosomal dominant 2, 600101 (3)	GJB3, CX31, DFNA2	603324	1p35.1	Ear,Nose,Throat
406	Deafness, autosomal dominant 2, 600101 (3)	KCNQ4, DFNA2	603537	1p34	Ear,Nose,Throat
406	Deafness, autosomal dominant 28, 608641 (3)	TFCP2L3, DFNA28	608576	8q22	Ear,Nose,Throat
406	Deafness, autosomal dominant 3, 601544 (3)	GJB2, CX26, DFNB1, PPK, DFNA3, KID, HID	121011	13q11-q12	Ear,Nose,Throat
406	Deafness, autosomal dominant 3, 601544 (3)	GJB6, CX30, DFNA3, HED, ED2	604418	13q12	Ear,Nose,Throat
406	Deafness, autosomal dominant 36, 606705 (3)	TMC1, DFNB7, DFNB11, DFNA36	606706	9q13-q21	Ear,Nose,Throat
406	Deafness, autosomal dominant 36, with dentinogenesis, 605594 (3)	DSPP, DPP, DGI1, DFNA39, DTDP2	125485	4q21.3	Ear,Nose,Throat
406	Deafness, autosomal dominant 40 (3)	CRYM, DFNA40	123740	16p13.11-p12.3	Ear,Nose,Throat
406	Deafness, autosomal dominant 4, 600652 (3)	MYH14, KIAA2034, DFNA4	608568	19q13.33	Ear,Nose,Throat
406	Deafness, autosomal dominant 5 (3)	DFNA5	600994	7p15	Ear,Nose,Throat
406	Deafness, autosomal dominant 8, 601543 (3)	TECTA, DFNA8, DFNA12, DFNB21	602574	11q22-q24	Ear,Nose,Throat
406	Deafness, autosomal dominant 9, 601369 (3)	COCH, DFNA9	603196	14q12-q13	Ear,Nose,Throat
406	Deafness, autosomal dominant nonsyndromic sensorineural, 607841 (3)	MYO1A	601478	12q13-q15	Ear,Nose,Throat
406	Deafness, autosomal dominant, with peripheral neuropathy (3)	GJB3, CX31, DFNA2	603324	1p35.1	Ear,Nose,Throat
406	Deafness, autosomal recessive 10, congenital, 605316 (3)	TMPRSS3, ECHOS1, DFNB8, DFNB10	605511	21q22.3	Ear,Nose,Throat
406	Deafness, autosomal recessive 1, 220290 (3)	GJB2, CX26, DFNB1, PPK, DFNA3, KID, HID	121011	13q11-q12	Ear,Nose,Throat
406	Deafness, autosomal recessive 12, 601386 (3)	CDH23, USH1D	605516	10q21-q22	Ear,Nose,Throat

406	Deafness, autosomal recessive 12, modifier of, 601386 (3)	ATP2B2, PMCA2	108733	3p26-p25	Ear,Nose,Throat
406	Deafness, autosomal recessive 16, 603720 (3)	STRC, DFNB16	606440	15q15	Ear,Nose,Throat
406	Deafness, autosomal recessive 18, 602092 (3)	USH1C, DFNB18	605242	11p15.1	Ear,Nose,Throat
406	Deafness, autosomal recessive 21, 603629 (3)	TECTA, DFNA8, DFNA12, DFNB21	602574	11q22-q24	Ear,Nose,Throat
406	Deafness, autosomal recessive 22, 607039 (3)	OTOA, DFNB22	607038	16p12.2	Ear,Nose,Throat
406	Deafness, autosomal recessive 23, 609533 (3)	PCDH15, DFNB23	605514	10q21-q22	Ear,Nose,Throat
406	Deafness, autosomal recessive 29 (3)	CLDN14, DFNB29	605608	21q22.3	Ear,Nose,Throat
406	Deafness, autosomal recessive 2, neurosensory, 600060 (3)	MYO7A, USH1B, DFNB2, DFNA11	276903	11q13.5	Ear,Nose,Throat
406	Deafness, autosomal recessive 30, 607101 (3)	MYO3A, DFNB30	606808	10p11.1	Ear,Nose,Throat
406	Deafness, autosomal recessive 31, 607084 (3)	WHRN, CIP98, KIAA1526, DFNB31	607928	9q32-q34	Ear,Nose,Throat
406	Deafness, autosomal recessive 3, 600316 (3)	MYO15A, DFNB3	602666	17p11.2	Ear,Nose,Throat
406	Deafness, autosomal recessive 36, 609006 (3)	ESPN	606351	1p36.3-p36.1	Ear,Nose,Throat
406	Deafness, autosomal recessive 37, 607821 (3)	MYO6, DFNA22, DFNB37	600970	6q13	Ear,Nose,Throat
406	Deafness, autosomal recessive (3)	GJB3, CX31, DFNA2	603324	1p35.1	Ear,Nose,Throat
406	Deafness, autosomal recessive 4, 600791 (3)	SLC26A4, PDS, DFNB4	605646	7q31	Ear,Nose,Throat
406	Deafness, autosomal recessive 61 (3)	PRES, DFNB61, SLC26A5	604943	7q22.1	Ear,Nose,Throat
406	Deafness, autosomal recessive 6, 600971 (3)	TMIE, DFNB6	607237	3p21	Ear,Nose,Throat
406	Deafness, autosomal recessive 7, 600974 (3)	TMC1, DFNB7, DFNB11, DFNA36	606706	9q13-q21	Ear,Nose,Throat
406	Deafness, autosomal recessive 8, childhood onset, 601072 (3)	TMPRSS3, ECHOS1, DFNB8, DFNB10	605511	21q22.3	Ear,Nose,Throat
406	Deafness, autosomal recessive 9, 601071 (3)	OTOF, DFNB9, NSRD9	603681	2p23-p22	Ear,Nose,Throat
406	Deafness, congenital heart defects, and posterior embryotoxon (3)	JAG1, AGS, AHD	601920	20p12	Ear,Nose,Throat
406	Deafness, nonsyndromic (3) ()	KIAA1199	608366	15q24	Ear,Nose,Throat
406	Deafness, nonsyndromic neurosensory, digenic (3)	GJB6, CX30, DFNA3, HED, ED2	604418	13q12	Ear,Nose,Throat
406	Deafness, sensorineural, with hypertrophic cardiomyopathy, 606346 (3)	MYO6, DFNA22, DFNB37	600970	6q13	Ear,Nose,Throat
406	Deafness, X-linked 1, progressive (3)	TIMM8A, DFN1, DDP, MTS, DDP1	300356	Xq22	Ear,Nose,Throat
406	Deafness, X-linked 3, conductive, with stapes fixation, 304400 (3)	POU3F4, DFN3	300039	Xq21.1	Ear,Nose,Throat
407	Debrisoquine sensitivity (3)	CYP2D@, CYP2D, P450C2D	124030	22q13.1	Metabolic
410	Dejerine-Sottas disease, 145900 (3)	PMP22, CMT1A, CMT1E, DSS	601097	17p11.2	multiple
410	Dejerine-Sottas neuropathy, 145900 (3)	EGR2, KROX20	129010	10q21.1-q22.1	multiple
410	Dejerine-Sottas neuropathy, autosomal recessive, 145900 (3)	PRX, CMT4F	605725	19q13.1-q13.2	multiple
410	Dejerine-Sottas syndrome, 145900 (3)	MPZ, CMT1B, CMTDI3, CHM, DSS	159440	1q22	multiple
411	Delayed sleep phase syndrome, susceptibility to (3)	AANAT, SNAT	600950	17q25	Psychiatric
412	Dementia, familial British, 176500 (3)	ITM2B, BRI, ABRI, FBD	603904	13q14	Neurological
412	Dementia, familial Danish, 117300 (3)	ITM2B, BRI, ABRI, FBD	603904	13q14	Neurological
412	Dementia, frontotemporal, 600274 (3)	PSEN1, AD3	104311	14q24.3	Neurological
412	Dementia, frontotemporal, with parkinsonism, 600274 (3)	MAPT, MTBT1, DDPAC, MSTD	157140	17q21.1	Neurological
412	Dementia, Lewy body, 127750 (3)	SNCA, NACP, PARK1, PARK4	163890	4q21	Neurological
412	Dementia, Lewy body, 127750 (3)	SNCB	602569	5q35	Neurological
412	Dementia, Pick disease-like, 172700 (3)	MAPT, MTBT1, DDPAC, MSTD	157140	17q21.1	Neurological
412	Dementia, vascular, susceptibility to (3)	TNF, TNFA	191160	6p21.3	Neurological
413	Dengue fever, protection against (3)	CD209, CDSIGN	604672	19p13.3	Immunological
414	Dental anomalies, isolated (3)	RUNX2, CBFA1, PEBP2A1, AML3	600211	6p21	Skeletal
415	Dentatorubro-pallidoluysian atrophy, 125370 (3)	DRPLA	607462	12p13.31	Neurological
416	Dent disease, 300009 (3)	CLCN5, CLCK2, NPHL2, DENTS	300008	Xp11.22	Renal
417	Dentin dysplasia, type II, 125420 (3)	DSPP, DPP, DGI1, DFNA39, DTDP2	125485	4q21.3	Bone
418	Dentinogenesis imperfecta, Shields type II, 125490 (3)	DSPP, DPP, DGI1, DFNA39, DTDP2	125485	4q21.3	Bone
418	Dentinogenesis imperfecta, Shields type III, 125500 (3)	DSPP, DPP, DGI1, DFNA39, DTDP2	125485	4q21.3	Bone
416	Dent syndrome, 300009 (3)	OCRL, LOCR, OCRL1, NPHL2	300535	Xq26.1	Renal
420	Denys-Drash syndrome, 194080 (3)	WT1	607102	11p13	Renal
422	Dermatofibrosarcoma protuberans (3)	PDGFB, SIS	190040	22q12.3-q13.1	Cancer

424	De Sanctis-Cacchione syndrome, 278800 (3)	ERCC6, CKN2, COFS, CSB	609413	10q11	multiple
425	Desmoid disease, hereditary, 135290 (3)	APC, GS, FPC	175100	5q21-q22	Cancer
426	Desmosterolosis, 602398 (3)	DHCR24, KIAA0018	606418	1p33-p31.1	Metabolic
2427	Diabetes insipidus, nephrogenic, 304800 (3)	AVPR2, DIR, DI1, ADHR	300538	Xq28	Endocrine
2427	Diabetes insipidus, nephrogenic, autosomal dominant, 125800 (3)	AQP2	107777	12q13	Endocrine
2427	Diabetes insipidus, nephrogenic, autosomal recessive, 222000 (3)	AQP2	107777	12q13	Endocrine
2427	Diabetes insipidus, neurohypophyseal, 125700 (3)	AVP, AVRP, VP	192340	20p13	Endocrine
427	Diabetes mellitus, 125853 (3)	ABCC8, SUR, PHHI, SUR1	600509	11p15.1	Endocrine
427	Diabetes mellitus, insulin-dependent, 222100 (3)	TCF1, HNF1A, MODY3	142410	12q24.2	Endocrine
427	Diabetes mellitus, insulin-dependent, 5, 600320 (3)	SUMO4, IDDM5	608829	6q25	Endocrine
427	Diabetes mellitus, insulin-dependent, susceptibility to, 222100 (3)	PTPN8, PEP, PTPN22, LYP	600716	1p13	Endocrine
427	Diabetes mellitus, insulin-resistant, with acanthosis nigricans (3)	INSR	147670	19p13.2	Endocrine
427	Diabetes mellitus, insulin-resistant, with acanthosis nigricans and hypertension, 604367 (3)	PPARG, PPARG1, PPARG2	601487	3p25	Endocrine
427	Diabetes mellitus, neonatal-onset, 606176 (3)	GCK	138079	7p15-p13	Endocrine
427	Diabetes mellitus, noninsulin-dependent, 125853 (3)	GCGR	138033	17q25	Endocrine
427	Diabetes mellitus, noninsulin-dependent, 125853 (3)	GPD2	138430	2q24.1	Endocrine
427	Diabetes mellitus, noninsulin-dependent, 125853 (3)	HNF4A, TCF14, MODY1	600281	20q12-q13.1	Endocrine
427	Diabetes mellitus, noninsulin-dependent, 125853 (3)	IRS2	600797	13q34	Endocrine
427	Diabetes mellitus, noninsulin-dependent, 125853 (3)	MAPK8IP1, IB1	604641	11p12-p11.2	Endocrine
427	Diabetes mellitus, noninsulin-dependent, 125853 (3)	NEUROD1, NIDDM	601724	2q32	Endocrine
427	Diabetes mellitus, noninsulin-dependent, 125853 (3)	TCF2, HNF2	189907	17cen-q21.3	Endocrine
427	Diabetes mellitus, noninsulin-dependent, 2, 125853 (3)	TCF1, HNF1A, MODY3	142410	12q24.2	Endocrine
427	Diabetes mellitus, noninsulin-dependent (3)	IRS1	147545	2q36	Endocrine
427	Diabetes mellitus, noninsulin-dependent (3)	SLC2A2, GLUT2	138160	3q26.1-q26.3	Endocrine
427	Diabetes mellitus, noninsulin-dependent (3)	SLC2A4, GLUT4	138190	17p13	Endocrine
427	Diabetes mellitus, noninsulin-dependent, 601283 (3)	CAPN10	605286	2q37.3	Endocrine
427	Diabetes mellitus, non-insulin-dependent, susceptibility to, 125853 (3)	ENPP1, PDNP1, NPPS, M6S1, PCA1	173335	6q22-q23	Endocrine
427	Diabetes mellitus, noninsulin-dependent, susceptibility to, 125853 (3)	RETN, RSTN, FIZZ3	605565	19p13.2	Endocrine
427	Diabetes mellitus, permanent neonatal, with cerebellar agenesis, 609069 (3)	PTF1A	607194	10p12.3	Endocrine
427	Diabetes mellitus, permanent neonatal, with neurologic features, 606176 (3)	KCNJ11, BIR, PHHI	600937	11p15.1	Endocrine
427	Diabetes mellitus, type II, 125853 (3)	AKT2	164731	19q13.1-q13.2	Endocrine
427	Diabetes mellitus, type II, susceptibility to, 125853 (3)	IPF1	600733	13q12.1	Endocrine
427	Diabetes mellitus, type I, susceptibility to, 222100 (3)	FOXP3, IPEX, AIID, XPID, PIDX	300292	Xp11.23-q13.3	Endocrine
427	Diabetes, permanent neonatal, 606176 (3)	KCNJ11, BIR, PHHI	600937	11p15.1	Endocrine
427	Diabetic nephropathy, susceptibility to, 603933 (3)	ACE, DCP1, ACE1	106180	17q23	Endocrine
427	Diabetic retinopathy, NIDDM-related, susceptibility to, 125853 (3)	VEGF	192240	6p12	Endocrine
430	Diastrophic dysplasia, 222600 (3)	SLC26A2, DTD, DTDST, D5S1708, EDM4	606718	5q32-q33.1	Skeletal
430	Diastrophic dysplasia, broad bone-platyspondylic variant (3)	SLC26A2, DTD, DTDST, D5S1708, EDM4	606718	5q32-q33.1	Skeletal
432	DiGeorge syndrome, 188400 (3)	TBX1, DGS, CTHM, CAFS, TGA, DORV, VCFS, DGCR	602054	22q11.2	multiple
433	Dihydropyrimidinuria (3)	DPYS, DHP	222748	8q22	Metabolic
434	Dilated cardiomyopathy with woolly hair and keratoderma, 605676 (3)	DSP, KPPS2, PPKS2	125647	6p24	multiple
435	Dimethylglycine dehydrogenase deficiency, 605850 (3)	DMGDH, DMGDHD	605849	5q12.2-q12.3	Metabolic
438	Disordered steroidogenesis, isolated (3)	POR	124015	7q11.2	Metabolic
439	Dissection of cervical arteries (3)	COL1A1	120150	17q21.31-q22	Connective tissue
2440	DNA ligase I deficiency (3)	LIG1	126391	19q13.2-q13.3	multiple
440	DNA topoisomerase I, camptothecin-resistant (3)	TOP1	126420	20q12-q13.1	Metabolic

440	DNA topoisomerase II, resistance to inhibition of, by amsacrine (3)	TOP2A, TOP2	126430	17q21-q22	Metabolic
441	Dopamine-beta-hydroxylase activity levels, plasma (3)	DBH	609312	9q34	Metabolic
441	Dopamine beta-hydroxylase deficiency, 223360 (3)	DBH	609312	9q34	Metabolic
443	Dosage-sensitive sex reversal, 300018 (3)	DAX1, AHC, AHX, NROB1	300473	Xp21.3-p21.2	Endocrine
444	Double-outlet right ventricle, 217095 (3)	CFC1, CRYPTIC, HTX2	605194	2q21.1	Cardiovascular
445	Down syndrome, risk of, 190685 (3)	MTR	156570	1q43	multiple
446	Doyne honeycomb degeneration of retina, 126600 (3)	EFEMP1, FBNL, DHRD	601548	2p16	Ophthamological
447	Drug addiction, susceptibility to (3)	FAAH	602935	1p35-p34	Psychiatric
449	Duane-radial ray syndrome, 607323 (3)	SALL4, HSAL4	607343	20q13.13-q13.2	multiple
450	Dubin-Johnson syndrome, 237500 (3)	ABCC2, CMOAT	601107	10q24	Metabolic
451	Duchenne muscular dystrophy, 310200 (3)	DMD, BMD	300377	Xp21.2	Muscular
452	Dyggve-Melchior-Clausen disease, 223800 (3)	DYM, FLJ90130, DMC, SMC	607461	18q12-q21.1	multiple
453	Dysalbuminemic hyperthyroxinemia (3)	ALB	103600	4q11-q13	Hematological
454	Dysautonomia, familial, 223900 (3)	IKBKAP, IKAP	603722	9q31	Neurological
455	Dyschromatosis symmetrica hereditaria, 127400 (3)	ADAR, DRADA, DSH, DSRAD	601059	1q21.3	Dermatological
456	Dyserythropoietic anemia with thrombocytopenia, 300367 (3)	GATA1, GF1, ERYF1, NFE1	305371	Xp11.23	Hematological
457	Dysfibrinogenemia, alpha type, causing bleeding diathesis (3)	FGA	134820	4q28	Hematological
457	Dysfibrinogenemia, alpha type, causing recurrent thrombosis (3)	FGA	134820	4q28	Hematological
457	Dysfibrinogenemia, beta type (3)	FGB	134830	4q28	Hematological
457	Dysfibrinogenemia, gamma type (3)	FGG	134850	4q28	Hematological
458	Dyskeratosis congenita-1, 305000 (3)	DKC1, DKC	300126	Xq28	Dermatological
458	Dyskeratosis congenita, autosomal dominant, 127550 (3)	TERC, TRC3, TR	602322	3q21-q28	Dermatological
459	Dyslexia, susceptibility to, 1, 127700 (3)	DYX1C1, DYXC1, DYX1	608706	15q21	Psychiatric
459	Dyslexia, susceptibility to, 2, 600202 (3)	KIAA0319, DYX2, DYLX2, DLX2	609269	6p22.2	Psychiatric
460	Dysprothrombinemia (3)	F2	176930	11p11-q12	Hematological
461	Dyssegmental dysplasia, Silverman-Handmaker type, 224410 (3)	HSPG2, PLC, SJS, SJA, SJS1	142461	1p36.1	Neurological
462	Dystonia-12, 128235 (3)	ATP1A3, DYT12, RDP	182350	19q12-q13.2	Neurological
462	Dystonia-1, torsion, 128100 (3)	DYT1, TOR1A	605204	9q34	Neurological
462	Dystonia, DOPA-responsive, 128230 (3)	GCH1, DYT5	600225	14q22.1-q22.2	Neurological
462	Dystonia, early-onset atypical, with myoclonic features (3)	DYT1, TOR1A	605204	9q34	Neurological
462	Dystonia, myoclonic, 159900 (3)	DRD2	126450	11q23	Neurological
462	Dystonia, myoclonic, 159900 (3)	SGCE, DYT11	604149	7q21	Neurological
462	Dystonia, primary cervical (3)	DRD5, DRD1B, DRD1L2	126453	4p16.1-p15.3	Neurological
463	Dystransthyretinemic hyperthyroxinemia(3)	TTR, PALB	176300	18q11.2-q12.1	Hematological
465	EBD, Bart type, 132000 (3)	COL7A1	120120	3p21.3	Dermatological
465	EBD, localisata variant (3)	COL7A1	120120	3p21.3	Dermatological
466	Ectodermal dysplasia-1, anhidrotic, 305100 (3)	ED1, EDA, HED	300451	Xq12-q13.1	Dermatological
466	Ectodermal dysplasia 2, hidrotic, 129500 (3)	GJB6, CX30, DFNA3, HED, ED2	604418	13q12	Dermatological
466	Ectodermal dysplasia, anhidrotic, 224900 (3)	EDARADD	606603	1q42.2-q43	Dermatological
466	Ectodermal dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 (3)	IKBKG, NEMO, FIP3, IP2	300248	Xq28	Dermatological
466	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency (3)	NFKBIA, IKBA	164008	14q13	Dermatological
466	Ectodermal dysplasia, hypohidrotic, autosomal dominant, 129490 (3)	EDAR, DL, ED3, EDA3	604095	2q11-q13	Dermatological
466	Ectodermal dysplasia, hypohidrotic, autosomal recessive, 224900 (3)	EDAR, DL, ED3, EDA3	604095	2q11-q13	Dermatological
466	Ectodermal dysplasia, hypohidrotic, with immune deficiency, 300291 (3)	IKBKG, NEMO, FIP3, IP2	300248	Xq28	Dermatological
466	Ectodermal dysplasia, Margarita Island type, 225060 (3)	HVEC, PVRL1, PVRR1, PRR1	600644	11q23-q24	Dermatological
466	Ectodermal dysplasia/skin fragility syndrome, 604536 (3)	PKP1	601975	1q32	Dermatological
467	Ectopia lentis, familial, 129600 (3)	FBN1, MFS1, WMS	134797	15q21.1	Ophthamological
467	Ectopia pupillae, 129750 (3)	PAX6, AN2, MGDA	607108	11p13	Ophthamological

468	Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 (3)	TP73L, TP63, KET, EEC3, SHFM4, LMS, RHS	603273	3q27	Skeletal
470	Ehlers-Danlos due to tenascin X deficiency, 606408 (3)	TNXB, TNX, TNXB1, TNXBS, TNXB2	600985	6p21.3	Connective tissue
470	Ehlers-Danlos syndrome, hypermobility type, 130020 (3)	TNXB, TNX, TNXB1, TNXBS, TNXB2	600985	6p21.3	Connective tissue
470	Ehlers-Danlos syndrome, progeroid form, 130070 (3)	B4GALT7, XGALT1, XGPT1	604327	5q35.2-q35.3	Connective tissue
470	Ehlers-Danlos syndrome, type I, 130000 (3)	COL1A1	120150	17q21.31-q22	Connective tissue
470	Ehlers-Danlos syndrome, type I, 130000 (3)	COL5A1	120215	9q34.2-q34.3	Connective tissue
470	Ehlers-Danlos syndrome, type I, 130000 (3)	COL5A2	120190	2q31	Connective tissue
470	Ehlers-Danlos syndrome, type II, 130010 (3)	COL5A1	120215	9q34.2-q34.3	Connective tissue
470	Ehlers-Danlos syndrome, type III, 130020 (3)	COL3A1	120180	2q31	Connective tissue
470	Ehlers-Danlos syndrome, type IV, 130050 (3)	COL3A1	120180	2q31	Connective tissue
470	Ehlers-Danlos syndrome, type VI, 225400 (3)	PLOD, PLOD1	153454	1p36.3-p36.2	Connective tissue
470	Ehlers-Danlos syndrome, type VII, 130060 (3)	COL1A1	120150	17q21.31-q22	Connective tissue
470	Ehlers-Danlos syndrome, type VIIA2, 130060 (3)	COL1A2	120160	7q22.1	Connective tissue
470	Ehlers-Danlos syndrome, type VIIC, 225410 (3)	ADAMTS2, NPI	604539	5q23	Connective tissue
471	Elite sprint athletic performance (3)	ACTN3	102574	11q13-q14	Muscular
472	Elliptocytosis-1 (3)	EPB41, EL1	130500	1p36.2-p34	Hematological
472	Elliptocytosis-2 (3)	SPTA1	182860	1q21	Hematological
472	Elliptocytosis-3 (3)	SPTB	182870	14q22-q23.2	Hematological
472	Elliptocytosis, Malaysian-Melanesian type (3)	SLC4A1, AE1, EPB3	109270	17q21-q22	Hematological
473	Ellis-van Creveld syndrome, 225500 (3)	EVC	604831	4p16	Skeletal
473	Ellis-van Creveld syndrome, 225500 (3)	LBN, EVC2	607261	4p16	Skeletal
474	Emery-Dreifuss muscular dystrophy, 310300 (3)	EMD, EDMD, STA	300384	Xq28	Muscular
474	Emery-Dreifuss muscular dystrophy, AD, 181350 (3)	LMNA, LMN1, EMD2, FPLD, CMD1A, HGPS, LGMD1B	150330	1q21.2	Muscular
474	Emery-Dreifuss muscular dystrophy, AR, 604929 (3)	LMNA, LMN1, EMD2, FPLD, CMD1A, HGPS, LGMD1B	150330	1q21.2	Muscular
475	Emphysema (3)	PI, AAT	107400	14q32.1	Respiratory
475	Emphysema-cirrhosis (3)	PI, AAT	107400	14q32.1	Respiratory
476	Encephalopathy, familial, with neuroserpin inclusion bodies, 604218 (3)	SERPINI1, PI12	602445	3q26	Neurological
476	Encephalopathy, progressive mitochondrial, with proximal renal tubulopathy due to cytochrome c oxidase deficiency (3)	COX10	602125	17p12-p11.2	Neurological
477	Enchondromatosis, Ollier type, 166000 (3)	PTHR1, PTHR	168468	3p22-p21.1	Cancer
479	Endometrial carcinoma (3)	CDH1, UVO	192090	16q22.1	Cancer
479	Endometrial carcinoma (3)	MSH3	600887	5q11-q12	Cancer
479	Endometrial carcinoma (3)	MSH6, GTBP, HNPCC5	600678	2p16	Cancer
479	Endometrial carcinoma (3)	PTEN, MMAC1	601728	10q23.31	Cancer
481	Endotoxin hyporesponsiveness (3)	TLR4	603030	9q32-q33	Immunological
482	Endplate acetylcholinesterase deficiency, 603034 (3)	COLQ, EAD	603033	3p25	Neurological
483	Enhanced S-cone syndrome, 268100 (3)	NR2E3, PNR, ESCS	604485	15q23	Ophthamological
484	Enlarged vestibular aqueduct, 603545 (3)	SLC26A4, PDS, DFNB4	605646	7q31	Ear,Nose,Throat
485	Enolase-beta deficiency (3)	ENO3	131370	17pter-p12	Metabolic
487	Enterokinase deficiency, 226200 (3)	PRSS7, ENTK	606635	21q21	Gastrointestinal
491	Eosinophil peroxidase deficiency, 261500 (3)	EPX	131399	17q23.1	Hematological
492	Epidermodysplasia verruciformis, 226400 (3)	EVER1, EV1	605828	17q25	Dermatological
492	Epidermodysplasia verruciformis, 226400 (3)	EVER2, EV2	605829	17q25	Dermatological
493	Epidermolysis bullosa dystrophica, AD, 131750 (3)	COL7A1	120120	3p21.3	Dermatological
493	Epidermolysis bullosa dystrophica, AR, 226600 (3)	COL7A1	120120	3p21.3	Dermatological
493	Epidermolysis bullosa, generalized atrophic benign, 226650 (3)	COL17A1, BPAG2	113811	10q24.3	Dermatological
493	Epidermolysis bullosa, generalized atrophic benign, 226650 (3)	ITGB4	147557	17q11-qter	Dermatological

493	Epidermolysis bullosa, generalized atrophic benign, 226650 (3)	LAMA3, LOCS	600805	18q11.2	Dermatological
493	Epidermolysis bullosa, generalized atrophic benign, 226650 (3)	LAMB3	150310	1q32	Dermatological
493	Epidermolysis bullosa, generalized atrophic benign, 226650 (3)	LAMC2, LAMNB2, LAMB2T	150292	1q25-q31	Dermatological
493	Epidermolysis bullosa, Herlitz junctional type, 226700 (3)	LAMB3	150310	1q32	Dermatological
493	Epidermolysis bullosa, Herlitz junctional type, 226700 (3)	LAMC2, LAMNB2, LAMB2T	150292	1q25-q31	Dermatological
493	Epidermolysis bullosa, junctional, Herlitz type, 226700 (3)	LAMA3, LOCS	600805	18q11.2	Dermatological
493	Epidermolysis bullosa, junctional, with pyloric atresia, 226730 (3)	ITGB4	147557	17q11-qter	Dermatological
493	Epidermolysis bullosa, junctional, with pyloric stenosis, 226730 (3)	ITGA6	147556	Chr.2	Dermatological
493	Epidermolysis bullosa, lethal acantholytic, 609638 (3)	DSP, KPPS2, PPKS2	125647	6p24	Dermatological
493	Epidermolysis bullosa of hands and feet, 131800 (3)	ITGB4	147557	17q11-qter	Dermatological
493	Epidermolysis bullosa, pretibial, 131850 (3)	COL7A1	120120	3p21.3	Dermatological
493	Epidermolysis bullosa pruriginosa, 604129 (3)	COL7A1	120120	3p21.3	Dermatological
493	Epidermolysis bullosa simplex, Koebner, Dowling-Meara, and Weber-Cockayne types, 131900, 131760, 131800 (3)	KRT14	148066	17q12-q21	Dermatological
493	Epidermolysis bullosa simplex, Koebner, Dowling-Meara, and Weber-Cockayne types, 131900, 131760, 131800 (3)	KRT5	148040	12q13	Dermatological
493	Epidermolysis bullosa simplex, Ogna type, 131950 (3)	PLEC1, PLTN, EBS1	601282	8q24	Dermatological
493	Epidermolysis bullosa simplex, recessive, 601001 (3)	KRT14	148066	17q12-q21	Dermatological
493	Epidermolysis bullosa simplex with mottled pigmentation, 131960 (3)	KRT5	148040	12q13	Dermatological
494	Epidermolytic hyperkeratosis, 113800 (3)	KRT10	148080	17q21-q22	Dermatological
494	Epidermolytic hyperkeratosis, 113800 (3)	KRT1	139350	12q13	Dermatological
494	Epidermolytic palmoplantar keratoderma, 144200 (3)	KRT9, EPPK	607606	17q12-q21	Dermatological
495	Epilepsy, benign, neonatal, type 1, 121200 (3)	KCNQ2, EBN1	602235	20q13.3	Neurological
495	Epilepsy, benign neonatal, type 2, 121201 (3)	KCNQ3, EBN2, BFNC2	602232	8q24	Neurological
495	Epilepsy, childhood absence, 607681 (3)	GABRG2, GEFSP3, CAE2, ECA2	137164	5q31.1-q33.1	Neurological
495	Epilepsy, childhood absence, 607682 (3)	CLCN2, EGMA, ECA3, EGI3	600570	3q26-qter	Neurological
495	Epilepsy, childhood absence, evolving to juvenile myoclonic epilepsy (3)	JRK, JH8	603210	8q24	Neurological
495	Epilepsy, generalized idiopathic, 600669 (3)	CACNB4, EJM	601949	2q22-q23	Neurological
495	Epilepsy, generalized, with febrile seizures plus, 604233 (3)	GABRG2, GEFSP3, CAE2, ECA2	137164	5q31.1-q33.1	Neurological
495	Epilepsy, generalized, with febrile seizures plus, type 2, 604233 (3)	SCN1A, GEFSP2, SMEI	182389	2q24	Neurological
495	Epilepsy, idopathic generalized, susceptibility to, 600669 (3)	ME2	154270	18q21	Neurological
495	Epilepsy, juvenile absence, 607631 (3)	CLCN2, EGMA, ECA3, EGI3	600570	3q26-qter	Neurological
495	Epilepsy, juvenile myoclonic, 606904 (3)	CACNB4, EJM	601949	2q22-q23	Neurological
495	Epilepsy, juvenile myoclonic, 606904 (3)	CLCN2, EGMA, ECA3, EGI3	600570	3q26-qter	Neurological
495	Epilepsy, juvenile myoclonic, 606904 (3)	GABRA1, EJM	137160	5q34-q35	Neurological
495	Epilepsy, myoclonic, Lafora type, 254780 (3)	EPM2A, MELF, EPM2	607566	6q24	Neurological
495	Epilepsy, myoclonic, Lafora type, 254780 (3)	NHLRC1, EPM2A, EPM2B	608072	6p22.3	Neurological
495	Epilepsy, neonatal myoclonic, with suppression-burst pattern, 609304 (3)	SLC25A22, GC1	609302	11p15.5	Neurological
495	Epilepsy, nocturnal frontal lobe, 1, 600513 (3)	CHRNA4, ENFL1	118504	20q13.2-q13.3	Neurological
495	Epilepsy, nocturnal frontal lobe, 3, 605375 (3)	CHRNB2, EFNL3	118507	1q21	Neurological
495	Epilepsy, partial, with auditory features, 600512 (3)	LGI1, EPT, ETL1	604619	10q24	Neurological
495	Epilepsy, progressive myoclonic 1, 254800 (3)	CSTB, STFB, EPM1	601145	21q22.3	Neurological
495	Epilepsy, progressive myoclonic 2B, 254780 (3)	NHLRC1, EPM2A, EPM2B	608072	6p22.3	Neurological
495	Epilepsy, severe myoclonic, of infancy, 607208 (3)	SCN1A, GEFSP2, SMEI	182389	2q24	Neurological
495	Epilepsy with grand mal seizures on awakening, 607628 (3)	CLCN2, EGMA, ECA3, EGI3	600570	3q26-qter	Neurological
495	Epilepsy, X-linked, with variable learning disabilities and behavior disorders, 300491 (3)	SYN1	313440	Xp11.4-p11.2	Neurological
496	Epiphyseal dysplasia, multiple 1, 132400 (3)	COMP, EDM1, MED, PSACH	600310	19p13.1	Bone
496	Epiphyseal dysplasia, multiple, 226900 (3)	SLC26A2, DTD, DTDST, D5S1708, EDM4	606718	5q32-q33.1	Bone

496	Epiphyseal dysplasia, multiple, 3, 600969 (3)	COL9A3, EDM3, IDD	120270	20q13.3	Bone
496	Epiphyseal dysplasia, multiple, 5, 607078 (3)	MATN3, EDM5, HOA	602109	2p24-p23	Bone
496	Epiphyseal dysplasia, multiple, COL9A1-related (3)	COL9A1, MED	120210	6q13	Bone
496	Epiphyseal dysplasia, multiple, type 2, 600204 (3)	COL9A2, EDM2	120260	1p33-p32.2	Bone
496	Epiphyseal dysplasia, multiple, with myopathy (3)	COL9A3, EDM3, IDD	120270	20q13.3	Bone
497	Episodic ataxia/myokymia syndrome, 160120 (3)	KCNA1, AEMK, EA1	176260	12p13	Neurological
497	Episodic ataxia, type 2, 108500 (3)	CACNA1A, CACNL1A4, SCA6	601011	19p13	Neurological
498	Epithelial ovarian cancer, somatic, 604370 (3)	OPCML	600632	11q25	Cancer
500	Epstein syndrome, 153650 (3)	MYH9, MHA, FTNS, DFNA17	160775	22q11.2	Hematological
502	Erythermalgia, primary, 133020 (3)	SCN9A, NENA, PN1	603415	2q24	Neurological
504	Erythremias, alpha- (3)	HBA1	141800	16pter-p13.3	Hematological
504	Erythremias, beta- (3)	HBB	141900	11p15.5	Hematological
505	Erythrocytosis (3)	HBA2	141850	16pter-p13.3	Hematological
505	Erythrocytosis, familial, 133100 (3)	EPOR	133171	19p13.3-p13.2	Hematological
507	Erythrokeratoderma, progressive symmetric, 602036 (3)	LOR	152445	1q21	Dermatological
507	Erythrokeratodermia variabilis, 133200 (3)	GJB3, CX31, DFNA2	603324	1p35.1	Dermatological
507	Erythrokeratodermia variabilis with erythema gyratum repens, 133200 (3)	GJB4, CX30.3	605425	1p35.1	Dermatological
508	Esophageal cancer, 133239 (3)	TGFBR2, HNPCC6	190182	3p22	Cancer
508	Esophageal carcinoma, somatic, 133239 (3)	RNF6	604242	13q12.11	Cancer
508	Esophageal squamous cell carcinoma, 133239 (3)	LZTS1, F37, FEZ1	606551	8p22	Cancer
508	Esophageal squamous cell carcinoma, 133239 (3)	WWOX, FOR	605131	16q23.3-q24.1	Cancer
509	Estrogen resistance (3)	ESR1, ESR	133430	6q25.1	Endocrine
510	Ethylmalonic encephalopathy, 602473 (3)	ETHE1, HSCO, D83198	608451	19q13.32	Metabolic
511	Ewing sarcoma (3)	EWSR1, EWS	133450	22q12	Cancer
512	Exertional myoglobinuria due to deficiency of LDH-A (3)	LDHA, LDH1	150000	11p15.4	Metabolic
514	Exostoses, multiple, type 1, 133700 (3)	EXT1	608177	8q24.11-q24.13	Bone
514	Exostoses, multiple, type 2, 133701 (3)	EXT2	608210	11p12-p11	Bone
515	Exudative vitreoretinopathy, 133780 (3)	FZD4, EVR1	604579	11q14-q21	Ophthamological
515	Exudative vitreoretinopathy, dominant, 133780 (3)	LRP5, BMND1, LRP7, LR3, OPPG, VBCH2	603506	11q13.4	Ophthamological
515	Exudative vitreoretinopathy, recessive, 601813 (3)	LRP5, BMND1, LRP7, LR3, OPPG, VBCH2	603506	11q13.4	Ophthamological
515	Exudative vitreoretinopathy, X-linked, 305390 (3)	NDP, ND	310600	Xp11.4	Ophthamological
516	Eye anomalies, multiplex (3)	PAX6, AN2, MGDA	607108	11p13	Ophthamological
517	Ezetimibe, nonresponse to (3)	NPC1L1	608010	7p13	Unclassified
518	Fabry disease (3)	GLA	301500	Xq22	Metabolic
519	Facioscapulohumeral muscular dystrophy-1A (3)	FSHMD1A, FSHD1A	158900	4q35	Muscular
520	Factor H and factor H-like 1 (3)	HF1, CFH, HUS	134370	1q32	Hematological
520	Factor V and factor VIII, combined deficiency of, 227300 (3)	MCFD2	607788	2p21-p16.3	Hematological
520	Factor VII deficiency (3)	F7	227500	13q34	Hematological
520	Factor X deficiency (3)	F10	227600	13q34	Hematological
520	Factor XI deficiency, autosomal dominant (3)	F11	264900	4q35	Hematological
520	Factor XI deficiency, autosomal recessive (3)	F11	264900	4q35	Hematological
520	Factor XII deficiency (3)	F12, HAF	234000	5q33-qter	Hematological
520	Factor XIIIA deficiency (3)	F13A1, F13A	134570	6p25-p24	Hematological
520	Factor XIIIB deficiency (3)	F13B	134580	1q31-q32.1	Hematological
522	Familial Mediterranean fever, 249100 (3)	MEFV, MEF, FMF	608107	16p13	Immunological
523	Fanconi anemia, complementation group A, 227650 (3)	FANCA, FACA, FA1, FA, FAA	607139	16q24.3	multiple
523	Fanconi anemia, complementation group B, 300514 (3)	FAAP95, FAAP90, FLJ34064, FANCB	300515	Xp22.31	multiple
523	Fanconi anemia, complementation group C (3)	FANCC, FACC	227645	9q22.3	multiple

523	Fanconi anemia, complementation group D1, 605724 (3)	BRCA2, FANCD1	600185	13q12.3	multiple
523	Fanconi anemia, complementation group D2 (3)	FANCD2, FANCD, FACD, FAD	227646	3p25.3	multiple
523	Fanconi anemia, complementation group E (3)	FANCE, FACE	600901	6p22-p21	multiple
523	Fanconi anemia, complementation group F (3)	FANCF	603467	11p15	multiple
523	Fanconi anemia, complementation group G (3)	XRCC9, FANCG	602956	9p13	multiple
523	Fanconi anemia, complementation group J, 609054 (3)	BRIP1, BACH1, FANCJ	605882	17q22	multiple
523	Fanconi anemia, complementation group L (3)	PHF9, FANCL	608111	2p16.1	multiple
523	Fanconi anemia, complementation group M (3)	FANCM, KIAA1596	609644	14q21.3	multiple
524	Fanconi-Bickel syndrome, 227810 (3)	SLC2A2, GLUT2	138160	3q26.1-q26.3	Metabolic
526	Farber lipogranulomatosis (3)	ASAH, AC	228000	8p22-p21.3	Metabolic
527	Fatty liver, acute, of pregnancy (3)	HADHA, MTPA	600890	2p23	Metabolic
528	Favism (3)	G6PD, G6PD1	305900	Xq28	Metabolic
530	Fechtner syndrome, 153640 (3)	MYH9, MHA, FTNS, DFNA17	160775	22q11.2	multiple
531	Feingold syndrome, 164280 (3)	MYCN, NMYC, ODED, MODED	164840	2p24.1	multiple
532	Fertile eunuch syndrome, 228300 (3)	GNRHR, LHRHR	138850	4q21.2	Endocrine
535	Fibrocalculous pancreatic diabetes, susceptibility to (3)	SPINK1, PSTI, PCTT, TATI	167790	5q32	Gastrointestinal
537	Fibromatosis, gingival, 135300 (3)	SOS1, GINGF, GF1, HGF	182530	2p22-p21	Connective tissue
537	Fibromatosis, juvenile hyaline, 228600 (3)	ANTXR2, CMG2, JHF, ISH	608041	4q21	Connective tissue
538	Fibrosis of extraocular muscles, congenital, 1, 135700 (3)	KIF21A, KIAA1708, FEOM1, CFEOM1	608283	12q12	Ophthamological
538	Fibrosis of extraocular muscles, congenital, 2, 602078 (3)	PHOX2A, ARIX, CFEOM2	602753	11q13.3-q13.4	Ophthamological
539	Fibular hypoplasia and complex brachydactyly, 228900 (3)	GDF5, CDMP1	601146	20q11.2	Skeletal
540	Fish-eye disease, 136120 (3)	LCAT	606967	16q22.1	Metabolic
541	Fish-odor syndrome, 602079 (3)	FMO3	136132	1q23-q25	Metabolic
542	Fitzgerald factor deficiency (3)	KNG	228960	3q27	Hematological
544	Fluorouracil toxicity, sensitivity to (3)	DPYD, DPD	274270	1p22	Metabolic
545	Focal cortical dysplasia, Taylor balloon cell type, 607341 (3)	TSC1, LAM	605284	9q34	Developmental
546	Follicle-stimulating hormone deficiency, isolated, 229070 (3)	FSHB	136530	11p13	Endocrine
547	Forebrain defects (3)	TDGF1	187395	3p23-p21	Neurological
548	Foveal hypoplasia, isolated, 136520 (3)	PAX6, AN2, MGDA	607108	11p13	Ophthamological
549	Foveomacular dystrophy, adult-onset, with choroidal neovascularization, 608161 (3)	RDS, RP7, PRPH2, PRPH, AVMD, AOFMD	179605	6p21.1-cen	Ophthamological
550	Fragile X syndrome (3)	FMR1, FRAXA	309550	Xq27.3	Neurological
551	Fraser syndrome, 219000 (3)	FRAS1	607830	4q21	multiple
551	Fraser syndrome, 219000 (3)	FREM2	608945	13q13.3	multiple
552	Frasier syndrome, 136680 (3)	WT1	607102	11p13	multiple
553	Friedreich ataxia, 229300 (3)	FRDA, FARR	606829	9q13	Neurological
553	Friedreich ataxia with retained reflexes, 229300 (3)	FRDA, FARR	606829	9q13	Neurological
554	Frontometaphyseal dysplasia, 304120 (3)	FLNA, FLN1, ABPX, NHBP, OPD1, OPD2, FMD, MNS	300017	Xq28	Skeletal
555	Fructose-bisphosphatase deficiency (3)	FBP1	229700	9q22.2-q22.3	Metabolic
556	Fructose intolerance (3)	ALDOB	229600	9q22.3	Metabolic
557	Fructosuria (3)	KHK	229800	2p23.3-p23.2	Metabolic
558	Fuchs endothelial corneal dystrophy, 136800 (3)	COL8A2, FECD, PPCD2	120252	1p34.3-p32.3	Ophthamological
559	Fucosidosis (3)	FUCA1	230000	1p34	Metabolic
560	Fucosyltransferase-6 deficiency (3)	FUT6	136836	19p13.3	Metabolic
561	Fumarase deficiency, 606812 (3)	FH	136850	1q42.1	Metabolic
562	Fundus albipunctatus, 136880 (3)	RDH5	601617	12q13-q14	Ophthamological
562	Fundus albipunctatus, 136880 (3)	RLBP1	180090	15q26	Ophthamological
562	Fundus flavimaculatus, 248200 (3)	ABCA4, ABCR, STGD1, FFM, RP19	601691	1p21-p13	Ophthamological
563	G6PD deficiency (3)	G6PD, G6PD1	305900	Xq28	Metabolic

564	GABA-transaminase deficiency (3)	ABAT, GABAT	137150	16p13.3	Metabolic
565	Galactokinase deficiency with cataracts, 230200 (3)	GALK1	604313	17q24	Metabolic
566	Galactose epimerase deficiency, 230350 (3)	GALE	606953	1p36-p35	Metabolic
567	Galactosemia, 230400 (3)	GALT	606999	9p13	Metabolic
568	Galactosialidosis (3)	PPGB, GSL, NGBE, GLB2, CTSA	256540	20q13.1	Metabolic
570	GAMT deficiency (3)	GAMT	601240	19p13.3	Metabolic
571	Gardner syndrome (3)	APC, GS, FPC	175100	5q21-q22	Cancer
572	Gastric cancer, 137215 (3)	APC, GS, FPC	175100	5q21-q22	Cancer
572	Gastric cancer, 137215 (3)	IRF1, MAR	147575	5q31.1	Cancer
572	Gastric cancer, familial diffuse, 137215 (3)	CDH1, UVO	192090	16q22.1	Cancer
572	Gastric cancer risk after H. pylori infection, 137215 (3)	IL1B	147720	2q14	Cancer
572	Gastric cancer risk after H. pylori infection, 137215 (3)	IL1RN	147679	2q14.2	Cancer
572	Gastric cancer, somatic, 137215 (3)	CASP10, MCH4, ALPS2	601762	2q33-q34	Cancer
572	Gastric cancer, somatic, 137215 (3)	ERBB2, NGL, NEU, HER2	164870	17q21.1	Cancer
572	Gastric cancer, somatic, 137215 (3)	FGFR2, BEK, CFD1, JWS	176943	10q26	Cancer
572	Gastric cancer, somatic, 137215 (3)	KLF6, COPEB, BCD1, ZF9	602053	10p15	Cancer
572	Gastric cancer, somatic, 137215 (3)	MUTYH	604933	1p34.3-p32.1	Cancer
574	Gastrointestinal stromal tumor, somatic, 606764 (3)	KIT, PBT	164920	4q12	Cancer
574	Gastrointestinal stromal tumor, somatic, 606764 (3)	PDGFRA	173490	4q12	Cancer
575	Gaucher disease, 230800 (3)	GBA	606463	1q21	Metabolic
575	Gaucher disease, variant form (3)	PSAP, SAP1	176801	10q22.1	Metabolic
575	Gaucher disease with cardiovascular calcification, 231005 (3)	GBA	606463	1q21	Metabolic
576	Gaze palsy, horizontal, with progressive scoliosis, 607313 (3)	ROBO3, RBIG1, RIG1, HGPPS	608630	11q23-q25	Neurological
578	Generalized epilepsy and paroxysmal dyskinesia, 609446 (3)	KCNMA1, SLO	600150	10q22.3	Neurological
578	Generalized epilepsy with febrile seizures plus, 604233 (3)	SCN1B, GEFSP1	600235	19q13.1	Neurological
580	Germ cell tumor (3)	BCL10	603517	1p22	Cancer
580	Germ cell tumors, 273300 (3)	KIT, PBT	164920	4q12	Cancer
581	Gerstmann-Straussler disease, 137440 (3)	PRNP, PRIP	176640	20pter-p12	Neurological
582	Giant axonal neuropathy-1, 256850 (3)	GAN, GAN1	605379	16q24.1	Neurological
583	Giant-cell fibroblastoma (3)	PDGFB, SIS	190040	22q12.3-q13.1	Cancer
2584	Giant cell hepatitis, neonatal, 231100 (3)	CYP7B1	603711	8q21.3	Gastrointestinal
584	Giant platelet disorder, isolated (3)	GP1BB	138720	22q11.2	Hematological
586	Gilbert syndrome, 143500 (3)	UGT1A1, UGT1, GNT1	191740	2q37	Metabolic
587	Gitelman syndrome, 263800 (3)	SLC12A3, NCCT, TSC	600968	16q13	Renal
588	Glanzmann thrombasthenia, type A, 273800 (3)	ITGA2B, GP2B, CD41B	607759	17q21.32	Hematological
588	Glanzmann thrombasthenia, type B (3)	ITGB3, GP3A	173470	17q21.32	Hematological
589	Glaucoma 1A, primary open angle, juvenile-onset, 137750 (3)	MYOC, TIGR, GLC1A, JOAG, GPOA	601652	1q24.3-q25.2	Ophthamological
589	Glaucoma 1A, primary open angle, recessive (3)	MYOC, TIGR, GLC1A, JOAG, GPOA	601652	1q24.3-q25.2	Ophthamological
589	Glaucoma 1E, primary open angle, adult-onset, 137760 (3)	OPTN, GLC1E, FIP2, HYPL, NRP	602432	10p15-p14	Ophthamological
589	Glaucoma 3A, primary congenital, 231300 (3)	CYP1B1, GLC3A	601771	2p22-p21	Ophthamological
589	Glaucoma, early-onset, digenic (3)	CYP1B1, GLC3A	601771	2p22-p21	Ophthamological
589	Glaucoma, early-onset, digenic (3)	MYOC, TIGR, GLC1A, JOAG, GPOA	601652	1q24.3-q25.2	Ophthamological
589	Glaucoma, normal tension, susceptibility to, 606657 (3)	OPA1, NTG, NPG	605290	3q28-q29	Ophthamological
589	Glaucoma, normal tension, susceptibility to, 606657 (3)	OPTN, GLC1E, FIP2, HYPL, NRP	602432	10p15-p14	Ophthamological
589	Glaucoma, primary open angle, adult-onset, 137760 (3)	CYP1B1, GLC3A	601771	2p22-p21	Ophthamological
589	Glaucoma, primary open angle, juvenile-onset, 137750 (3)	CYP1B1, GLC3A	601771	2p22-p21	Ophthamological
590	Glioblastoma, early-onset, 137800 (3)	MSH2, COCA1, FCC1, HNPCC1	609309	2p22-p21	Cancer
590	Glioblastoma multiforme, somatic, 137800 (3)	DMBT1	601969	10q25.3-q26.1	Cancer
590	Glioblastoma, somatic, 137800 (3)	ERBB2, NGL, NEU, HER2	164870	17q21.1	Cancer
590	Glioblastoma, somatic, 137800 (3)	LGI1, EPT, ETL1	604619	10q24	Cancer

590	Glioblastoma, susceptibility to, 137800 (3)	PPARG, PPARG1, PPARG2	601487	3p25	Cancer
594	Glomerulocystic kidney disease, hypoplastic, 137920 (3)	TCF2, HNF2	189907	17cen-q21.3	Renal
596	Glomerulosclerosis, focal segmental, 1, 603278 (3)	ACTN4, FSGS1, FSGS	604638	19q13	Renal
596	Glomerulosclerosis, focal segmental, 2, 603965 (3)	TRPC6, TRP6, FSGS2	603652	11q21-q22	Renal
596	Glomerulosclerosis, focal segmental, 3, 607832 (3)	CD2AP, CMS	604241	Chr.6	Renal
597	Glomuvenous malformations, 138000 (3)	GLML, GVM, VMGLOM	601749	1p22-p21	Cardiovascular
598	Glucocorticoid deficiency 2, 607398 (3)	MRAP, FALP, C21orf61	609196	21q22.1	Endocrine
598	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200 (3)	MC2R	607397	18p11.2	Endocrine
599	Glucose/galactose malabsorption, 606824 (3)	SLC5A1, SGLT1	182380	22q13.1	Metabolic
600	Glucose transport defect, blood-brain barrier, 606777 (3)	SLC2A1, GLUT1	138140	1p35-p31.3	Metabolic
601	Glucosidase I deficiency, 606056 (3)	GCS1	601336	2p13-p12	Metabolic
602	Glutamate formiminotransferase deficiency, 229100 (3)	FTCD	606806	21q22.3	Metabolic
603	Glutaricaciduria, type I, 231670 (3)	GCDH	608801	19p13.2	Metabolic
603	Glutaricaciduria, type IIA, 231680 (3)	ETFA, GA2, MADD	608053	15q23-q25	Metabolic
603	Glutaricaciduria, type IIB, 231680 (3)	ETFB, MADD	130410	19q13.3	Metabolic
603	Glutaricaciduria, type IIC, 231680 (3)	ETFDH, MADD	231675	4q32-qter	Metabolic
604	Glutathione synthetase deficiency, 266130 (3)	GSS, GSHS	601002	20q11.2	Metabolic
607	Glycerol kinase deficiency, 307030 (3)	GK	300474	Xp21.3-p21.2	Metabolic
608	Glycine encephalopathy, 605899 (3)	AMT, NKH, GCE	238310	3p21.2-p21.1	Metabolic
608	Glycine encephalopathy, 605899 (3)	GCSH, NKH	238330	16q24	Metabolic
608	Glycine encephalopathy, 605899 (3)	GLDC, HYGN1, GCSP, GCE, NKH	238300	9p22	Metabolic
608	Glycine N-methyltransferase deficiency, 606664 (3)	GNMT	606628	6p12	Metabolic
609	Glycogenosis, hepatic, autosomal (3)	PHKG2	172471	16p12.1-p11.2	Metabolic
609	Glycogenosis, X-linked hepatic, type I (3)	PHKA2, PHK	306000	Xp22.2-p22.1	Metabolic
609	Glycogenosis, X-linked hepatic, type II (3)	PHKA2, PHK	306000	Xp22.2-p22.1	Metabolic
610	Glycogen storage disease I (3)	G6PC, G6PT	232200	17q21	Metabolic
610	Glycogen storage disease Ib, 232220 (3)	G6PT1	602671	11q23	Metabolic
610	Glycogen storage disease Ic, 232240 (3)	G6PT1	602671	11q23	Metabolic
610	Glycogen storage disease II, 232300 (3)	GAA	606800	17q25.2-q25.3	Metabolic
610	Glycogen storage disease IIb, 300257 (3)	LAMP2, LAMPB	309060	Xq24	Metabolic
610	Glycogen storage disease IIIa (3)	AGL, GDE	232400	1p21	Metabolic
610	Glycogen storage disease IIIb (3)	AGL, GDE	232400	1p21	Metabolic
610	Glycogen storage disease IV, 232500 (3)	GBE1	607839	3p12	Metabolic
610	Glycogen storage disease, type 0, 240600 (3)	GYS2	138571	12p12.2	Metabolic
610	Glycogen storage disease VI (3)	PYGL	232700	14q21-q22	Metabolic
610	Glycogen storage disease VII (3)	PFKM	232800	12q13.3	Metabolic
614	GM1-gangliosidosis (3)	GLB1	230500	3p21.33	Metabolic
614	GM2-gangliosidosis, AB variant (3)	GM2A	272750	5q31.3-q33.1	Metabolic
614	GM2-gangliosidosis, several forms, 272800 (3)	HEXA, TSD	606869	15q23-q24	Metabolic
615	Gnthodiaphyseal dysplasia, 166260 (3)	TMEM16E, GDD1	608662	11p14.3	Bone
617	Goiter, congenital (3)	TPO, TPX	606765	2p25	Endocrine
617	Goiter, nonendemic, simple (3)	TG, AITD3	188450	8q24.2-q24.3	Endocrine
618	Goldberg-Shprintzen megacolon syndrome, 609460 (3)	KIAA1279	609367	10q22.1	multiple
619	Gonadal dysgenesis, 46XY, partial, with minifascicular neuropathy, 607080 (3)	DHH	605423	12q13.1	Endocrine
619	Gonadal dysgenesis, XY type (3)	SRY, TDF	480000	Yp11.3	Endocrine
622	GRACILE syndrome, 603358 (3)	BCS1L, FLNMS, GRACILE	603647	2q33	Metabolic
623	Graft-versus-host disease, protection against (3)	IL10, CSIF	124092	1q31-q32	Immunological
624	Graves disease, susceptibility to, 275000 (3)	CTLA4	123890	2q33	Endocrine
624	Graves disease, susceptibility to, 3, 275000 (3)	GC, DBP	139200	4q12	Endocrine

625	Greenberg dysplasia, 215140 (3)	LBR, PHA	600024	1q42.1	Skeletal
626	Greig cephalopolysyndactyly syndrome, 175700 (3)	GLI3, PAPA, PAPB, ACLS	165240	7p13	Skeletal
627	Griscelli syndrome, type 1, 214450 (3)	MYO5A, MYH12, GS1	160777	15q21	Dermatological
627	Griscelli syndrome, type 2, 607624 (3)	RAB27A, RAM, GS2	603868	15q21	Dermatological
627	Griscelli syndrome, type 3, 609227 (3)	MLPH	606526	2q37	Dermatological
628	Growth hormone deficient dwarfism (3)	GHRHR	139191	7p15-p14	Endocrine
628	Growth hormone insensitivity with immunodeficiency, 245590 (3)	STAT5B	604260	17q11.2	Endocrine
2628	Growth retardation with deafness and mental retardation due to IGF1 deficiency, 608747 (3)	IGF1	147440	12q22-q24.1	Developmental
630	Guttmacher syndrome, 176305 (3)	HOXA13, HOX1J	142959	7p15-p14.2	multiple
632	Gyrate atrophy of choroid and retina with ornithinemia, B6 responsive or unresponsive (3)	OAT	258870	10q26	Metabolic
633	Hailey-Hailey disease, 169600 (3)	ATP2C1, BCPM, HHD	604384	3q21-q24	Dermatological
634	Haim-Munk syndrome, 245010 (3)	CTSC, CPPI, PALS, PLS, HMS	602365	11q14.1-q14.3	multiple
638	Hand-foot-uterus syndrome, 140000 (3)	HOXA13, HOX1J	142959	7p15-p14.2	multiple
639	Harderoporphyrinuria (3)	CPO	121300	3q12	Metabolic
640	HARP syndrome, 607236 (3)	PANK2, NBIA1, PKAN, HARP	606157	20p13-p12.3	Metabolic
641	Hartnup disorder, 234500 (3)	SLC6A19, HND	608893	5p15	Metabolic
643	Hay-Wells syndrome, 106260 (3)	TP73L, TP63, KET, EEC3, SHFM4, LMS, RHS	603273	3q27	multiple
644	HDL deficiency, familial, 604091 (3)	ABCA1, ABC1, HDLDT1, TGD	600046	9q22-q31	Metabolic
644	HDL response to hormone replacement, augmented (3)	ESR1, ESR	133430	6q25.1	Metabolic
646	Hearing loss, low-frequency sensorineural, 600965 (3)	WFS1, WFRS, WFS, DFNA6	606201	4p16.1	Ear,Nose,Throat
647	Heart block, nonprogressive, 113900 (3)	SCN5A, LQT3, IVF, HB1, SSS1	600163	3p21	Cardiovascular
647	Heart block, progressive, type I, 113900 (3)	SCN5A, LQT3, IVF, HB1, SSS1	600163	3p21	Cardiovascular
648	Heinz body anemia (3)	HBA2	141850	16pter-p13.3	Hematological
648	Heinz body anemias, alpha- (3)	HBA1	141800	16pter-p13.3	Hematological
648	Heinz body anemias, beta- (3)	HBB	141900	11p15.5	Hematological
649	HELLP syndrome, maternal, of pregnancy (3)	HADHA, MTPA	600890	2p23	Metabolic
650	Hemangioblastoma, cerebellar, somatic (3)	VHL	608537	3p26-p25	Cancer
651	Hemangioma, capillary infantile, somatic, 602089 (3)	FLT4, VEGFR3, PCL	136352	5q35.3	Cancer
651	Hemangioma, capillary infantile, somatic, 602089 (3)	KDR	191306	4q12	Cancer
652	Hematopoiesis, cyclic, 162800 (3)	ELA2	130130	19p13.3	Hematological
653	Hematuria, familial benign (3)	COL4A4	120131	2q36-q37	Renal
654	Heme oxygenase-1 deficiency (3)	HMOX1	141250	22q12	Metabolic
656	Hemiplegic migraine, familial, 141500 (3)	CACNA1A, CACNL1A4, SCA6	601011	19p13	Neurological
657	Hemochromatosis (3)	HFE, HLA-H, HFE1	235200	6p21.3	Metabolic
657	Hemochromatosis, juvenile, 602390 (3)	HAMP, LEAP1, HEPC, HFE2	606464	19q13	Metabolic
657	Hemochromatosis, juvenile, digenic, 602390 (3)	HAMP, LEAP1, HEPC, HFE2	606464	19q13	Metabolic
657	Hemochromatosis, type 2A, 602390 (3)	HJV, HFE2A	608374	1q21	Metabolic
657	Hemochromatosis, type 3, 604250 (3)	TFR2, HFE3	604720	7q22	Metabolic
657	Hemochromatosis, type 4, 606069 (3)	SLC40A1, SLC11A3, FPN1, IREG1, HFE4	604653	2q32	Metabolic
659	Hemoglobin H disease (3)	HBA2	141850	16pter-p13.3	Hematological
660	Hemolytic anemia due to adenylate kinase deficiency (3)	AK1	103000	9q34.1	Hematological
660	Hemolytic anemia due to band 3 defect defect (3)	SLC4A1, AE1, EPB3	109270	17q21-q22	Hematological
660	Hemolytic anemia due to bisphosphoglycerate mutase deficiency (3)	BPGM	222800	7q31-q34	Hematological
660	Hemolytic anemia due to G6PD deficiency (3)	G6PD, G6PD1	305900	Xq28	Hematological
660	Hemolytic anemia due to gamma-glutamylcysteine synthetase deficiency, 230450 (3)	GCLC, GLCLC	606857	6p12	Hematological

660	Hemolytic anemia due to glucosephosphate isomerase deficiency (3)	GPI	172400	19q13.1	Hematological
660	Hemolytic anemia due to glutathione synthetase deficiency, 231900 (3)	GSS, GSHS	601002	20q11.2	Hematological
660	Hemolytic anemia due to hexokinase deficiency (3)	HK1	142600	10q22	Hematological
660	Hemolytic anemia due to PGK deficiency (3)	PGK1, PGKA	311800	Xq13	Hematological
660	Hemolytic anemia due to triosephosphate isomerase deficiency (3)	TPI1	190450	12p13	Hematological
661	Hemolytic-uremic syndrome, 235400 (3)	HF1, CFH, HUS	134370	1q32	Hematological
662	Hemophagocytic lymphohistiocytosis, familial, 2, 603553 (3)	PRF1, HPLH2	170280	10q22	Hematological
662	Hemophagocytic lymphohistiocytosis, familial, 3, 608898 (3)	UNC13D, MUNC13-4, HPLH3, HLH3, FHL3	608897	17q25.1	Hematological
663	Hemophilia A (3)	F8, F8C, HEMA	306700	Xq28	Hematological
663	Hemophilia B (3)	F9, HEMB	306900	Xq27.1-q27.2	Hematological
664	Hemorrhagic diathesis due to \`antithrombin\' Pittsburgh (3)	PI, AAT	107400	14q32.1	Hematological
664	Hemorrhagic diathesis due to factor V deficiency (3)	F5	227400	1q23	Hematological
665	Hemosiderosis, systemic, due to aceruloplasminemia, 604290 (3)	CP	117700	3q23-q24	Hematological
668	Hepatic adenoma, 142330 (3)	TCF1, HNF1A, MODY3	142410	12q24.2	Cancer
666	Hepatic failure, early onset, and neurologic disorder (3)	SCOD1, SCO1	603644	17p13-p12	Gastrointestinal
4666	Hepatic lipase deficiency (3)	LIPC	151670	15q21-q23	Metabolic
668	Hepatoblastoma (3)	CTNNB1	116806	3p22-p21.3	Cancer
668	Hepatocellular cancer, 114550 (3)	PDGFRL, PDGRL, PRLTS	604584	8p22-p21.3	Cancer
668	Hepatocellular carcinoma, 114550 (3)	AXIN1, AXIN	603816	16p13.3	Cancer
668	Hepatocellular carcinoma, 114550 (3)	CTNNB1	116806	3p22-p21.3	Cancer
668	Hepatocellular carcinoma, 114550 (3)	TP53, P53, LFS1	191170	17p13.1	Cancer
668	Hepatocellular carcinoma (3)	IGF2R, MPRI	147280	6q26	Cancer
668	Hepatocellular carcinoma, childhood type, 114550 (3)	MET	164860	7q31	Cancer
668	Hepatocellular carcinoma, somatic, 114550 (3)	CASP8, MCH5	601763	2q33-q34	Cancer
669	Hereditary hemorrhagic telangiectasia-1, 187300 (3)	ENG, END, HHT1, ORW	131195	9q34.1	Cardiovascular
669	Hereditary hemorrhagic telangiectasia-2, 600376 (3)	ACVRL1, ACVRLK1, ALK1, HHT2	601284	12q11-q14	Cardiovascular
2669	Hereditary persistence of alpha-fetoprotein (3)	AFP, HPAFP	104150	4q11-q13	Hematological
670	Hermansky-Pudlak syndrome, 203300 (3)	HPS1	604982	10q23.1	multiple
670	Hermansky-Pudlak syndrome, 203300 (3)	HPS3	606118	3q24	multiple
670	Hermansky-Pudlak syndrome, 203300 (3)	HPS4	606682	22q11.2-q12.2	multiple
670	Hermansky-pudlak syndrome, 203300 (3)	HPS5, RU2, KIAA1017	607521	11p15-p13	multiple
670	Hermansky-Pudlak syndrome, 203300 (3)	HPS6, RU	607522	10q24.32	multiple
670	Hermansky-Pudlak syndrome, 608233 (3)	AP3B1, ADTB3A, HPS2	603401	5q14.1	multiple
670	Hermansky-Pudlak syndrome 7, 203300 (3)	DTNBP1, HPS7	607145	6p22.3	multiple
675	Heterotaxy, visceral, 605376 (3)	CFC1, CRYPTIC, HTX2	605194	2q21.1	multiple
675	Heterotaxy, X-linked visceral, 306955 (3)	ZIC3, HTX1, HTX	300265	Xq26.2	multiple
676	Heterotopia, periventricular, 300049 (3)	FLNA, FLN1, ABPX, NHBP, OPD1, OPD2, FMD, MNS	300017	Xq28	Neurological
676	Heterotopia, periventricular, ED variant, 300537 (3)	FLNA, FLN1, ABPX, NHBP, OPD1, OPD2, FMD, MNS	300017	Xq28	Neurological
676	Heterotopia, periventricular nodular, with frontometaphyseal dysplasia, 300049 (3)	FLNA, FLN1, ABPX, NHBP, OPD1, OPD2, FMD, MNS	300017	Xq28	Neurological
677	Hex A pseudodeficiency, 272800 (3)	HEXA, TSD	606869	15q23-q24	Metabolic
679	High-molecular-weight kininogen deficiency (3)	KNG	228960	3q27	Hematological
681	Hirschsprung disease, 142623 (3)	EDN3	131242	20q13.2-q13.3	Gastrointestinal
681	Hirschsprung disease, 142623 (3)	GDNF	600837	5p13.1-p12	Gastrointestinal
681	Hirschsprung disease, 142623 (3)	NRTN, NTN	602018	19p13.3	Gastrointestinal
681	Hirschsprung disease, 142623 (3)	RET, MEN2A	164761	10q11.2	Gastrointestinal
681	Hirschsprung disease-2, 600155 (3)	EDNRB, HSCR2, ABCDS	131244	13q22	Gastrointestinal

681	Hirschsprung disease, cardiac defects, and autonomic dysfunction (3)	ECE1	600423	1p36.1	Gastrointestinal
681	Hirschsprung disease, short-segment, 142623 (3)	PMX2B, NBPHOX, PHOX2B	603851	4p12	Gastrointestinal
682	Histidinemia, 235800 (3)	HAL, HSTD	609457	12q22-q23	Metabolic
683	Histiocytoma (3)	TP53, P53, LFS1	191170	17p13.1	Cancer
684	HIV-1 disease, delayed progression of (3)	CCL5, SCYA5, D17S136E, TCP228	187011	17q11.2-q12	Immunological
684	HIV-1 disease, rapid progression of (3)	CCL5, SCYA5, D17S136E, TCP228	187011	17q11.2-q12	Immunological
684	HIV-1, susceptibility to (3)	IL10, CSIF	124092	1q31-q32	Immunological
684	HIV infection, susceptibility/resistance to (3)	CMKBR2, CCR2	601267	3p21	Immunological
684	HIV infection, susceptibility/resistance to (3)	CMKBR5, CCCKR5	601373	3p21	Immunological
686	HMG-CoA lyase deficiency (3)	HMGCL	246450	1pter-p33	Metabolic
686	HMG-CoA synthase-2 deficiency, 605911 (3)	HMGCS2	600234	1p13-p12	Metabolic
688	Holocarboxylase synthetase deficiency, 253270 (3)	HLCS, HCS	609018	21q22.1	Metabolic
689	Holoprosencephaly-2, 157170 (3)	SIX3, HPE2	603714	2p21	Developmental
689	Holoprosencephaly-3, 142945 (3)	SHH, HPE3, HLP3, SMMCI	600725	7q36	Developmental
689	Holoprosencephaly-4, 142946 (3)	TGIF, HPE4	602630	18p11.3	Developmental
689	Holoprosencephaly-5, 609637 (3)	ZIC2, HPE5	603073	13q32	Developmental
689	Holoprosencephaly-7 (3)	PTCH, NBCCS, BCNS, HPE7	601309	9q22.3	Developmental
696	Holt-Oram syndrome, 142900 (3)	TBX5	601620	12q24.1	Developmental
697	Homocysteine, total plasma, elevated (3)	CTH	607657	1p31.1	Metabolic
698	Homocystinuria, B6-responsive and nonresponsive types (3)	CBS	236200	21q22.3	Metabolic
698	Homocystinuria due to MTHFR deficiency, 236250 (3)	MTHFR	607093	1p36.3	Metabolic
699	Homocystinuria-megaloblastic anemia, cbl E type, 236270 (3)	MTRR	602568	5p15.3-p15.2	Metabolic
701	Homozygous 2p16 deletion syndrome, 606407 (3)	SLC3A1, ATR1, D2H, NBAT	104614	2p16.3	multiple
702	Hoyeraal-Hreidarsson syndrome, 300240 (3)	DKC1, DKC	300126	Xq28	multiple
703	HPFH, deletion type (3)	HBB	141900	11p15.5	Hematological
703	HPFH, nondeletion type A (3)	HBG1	142200	11p15.5	Hematological
703	HPFH, nondeletion type G (3)	HBG2	142250	11p15.5	Hematological
704	HPRT-related gout, 300323 (3)	HPRT1, HPRT	308000	Xq26-q27.2	Metabolic
705	H. pylori infection, susceptibility to, 600263 (3)	IFNGR1	107470	6q23-q24	Immunological
708	Huntington disease (3)	HD, IT15	143100	4p16.3	Neurological
708	Huntington disease-like 1, 603218 (3)	PRNP, PRIP	176640	20pter-p12	Neurological
708	Huntington disease-like 2, 606438 (3)	JPH3, JP3, HDL2	605268	16q24.3	Neurological
708	Huntington disease-like-4, 607136 (3)	TBP, SCA17	600075	6q27	Neurological
710	Hyalinosis, infantile systemic, 236490 (3)	ANTXR2, CMG2, JHF, ISH	608041	4q21	multiple
712	Hydrocephalus due to aqueductal stenosis, 307000 (3)	L1CAM, CAML1, HSAS1	308840	Xq28	Neurological
712	Hydrocephalus with congenital idiopathic intestinal pseudoobstruction, 307000 (3)	L1CAM, CAML1, HSAS1	308840	Xq28	Neurological
712	Hydrocephalus with Hirschsprung disease and cleft palate, 142623 (3)	L1CAM, CAML1, HSAS1	308840	Xq28	Neurological
716	Hyperalphalipoproteinemia, 143470 (3)	CETP	118470	16q21	Metabolic
717	Hyperammonemia with hypoornithinemia, hypocitrullinemia, hypoargininemia, and hypoprolinemia (3)	PYCS, GSAS	138250	10q24.3	Metabolic
718	Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency (3)	CYP21A2, CYP21, CA21H	201910	6p21.3	Endocrine
719	Hyperapobetalipoproteinemia, susceptibility to (3)	PPARA, PPAR	170998	22q12-q13.1	Metabolic
720	Hyperbilirubinemia, familial transcient neonatal, 237900 (3)	UGT1A1, UGT1, GNT1	191740	2q37	Gastrointestinal
721	Hypercalciuria, absorptive, susceptibility to, 143870 (3)	SAC, HCA2	605205	1q24	Renal
723	Hypercholanemia, familial, 607748 (3)	BAAT	602938	9q22.3	Gastrointestinal
723	Hypercholanemia, familial, 607748 (3)	EPHX1	132810	1q42.1	Gastrointestinal
723	Hypercholanemia, familial, 607748 (3)	TJP2, ZO2	607709	9q12-q13	Gastrointestinal
724	Hypercholesterolemia, due to ligand-defective apo B, 144010 (3)	APOB, FLDB	107730	2p24	Metabolic
724	Hypercholesterolemia, familial, 143890 (3)	LDLR, FHC, FH	606945	19p13.2	Metabolic

724	Hypercholesterolemia, familial, 3, 603776 (3)	PCSK9, NARC1, HCHOLA3, FH3	607786	1p34.1-p32	Metabolic
724	Hypercholesterolemia, familial, autosomal recessive, 603813 (3)	ARH, FHCB2, FHCB1	605747	1p36-p35	Metabolic
724	Hypercholesterolemia, familial, due to LDLR defect, modifier of, 143890 (3)	EPHX2	132811	8p21-p12	Metabolic
724	Hypercholesterolemia, familial, modification of, 143890 (3)	APOA2	107670	1q21-q23	Metabolic
724	Hypercholesterolemia, susceptibility to, 143890 (3)	GSBS	604088	7p15	Metabolic
724	Hypercholesterolemia, susceptibility to, 143890 (3)	ITIH4, PK120, ITIHL1	600564	3p21.2-p14.1	Metabolic
725	Hyperekplexia and spastic paraparesis (3)	GLRA1, STHE	138491	5q32	Neurological
725	Hyperekplexia, autosomal recessive, 149400 (3)	GLRB	138492	4q31.3	Neurological
726	Hypereosinophilic syndrome, idiopathic, resistant to imatinib, 607685 (3)	PDGFRA	173490	4q12	Hematological
727	Hyperferritinemia-cataract syndrome, 600886 (3)	FTL	134790	19q13.3-q13.4	Ophthamological
728	Hyper-IgD syndrome, 260920 (3)	MVK, MVLK	251170	12q24	Immunological
731	Hyperinsulinism, familial, 602485 (3)	GCK	138079	7p15-p13	Metabolic
732	Hyperinsulinism-hyperammonemia syndrome, 606762 (3)	GLUD1	138130	10q23.3	Metabolic
733	Hyperkalemic periodic paralysis, 170500 (3)	SCN4A, HYPP, NAC1A	603967	17q23.1-q25.3	Neurological
734	Hyperkeratotic cutaneous capillary-venous malformations associated with cerebral capillary malformations, 116860 (3)	CCM1, CAM, KRIT1	604214	7q11.2-q21	Neurological
736	Hyperlipidemia, familial combined, susceptibility to, 602491 (3)	USF1, HYPLIP1	191523	1q22-q23	Metabolic
737	Hyperlipoproteinemia, type Ib, 207750 (3)	APOC2	608083	19q13.2	Metabolic
737	Hyperlipoproteinemia, type III (3)	APOE, AD2	107741	19q13.2	Metabolic
738	Hyperlysinemia, 238700 (3)	AASS	605113	7q31.3	Metabolic
739	Hypermethioninemia, persistent, autosomal dominant, due to methionine adenosyltransferase I/III deficiency (3)	MAT1A, MATA1, SAMS1	250850	10q22	Metabolic
739	Hypermethioninemia with deficiency of S-adenosylhomocysteine hydrolase (3)	AHCY, SAHH	180960	20cen-q13.1	Metabolic
740	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome, 238970 (3)	SLC25A15, ORNT1, HHH	603861	13q14	Metabolic
741	Hyperostosis, endosteal, 144750 (3)	LRP5, BMND1, LRP7, LR3, OPPG, VBCH2	603506	11q13.4	Bone
742	Hyperoxaluria, primary, type 1, 259900 (3)	AGXT, SPAT	604285	2q36-q37	Metabolic
742	Hyperoxaluria, primary, type II, 260000 (3)	GRHPR, GLXR	604296	9cen	Metabolic
743	Hyperparathyroidism, AD, 145000 (3)	MEN1	131100	11q13	Endocrine
743	Hyperparathyroidism, familial primary, 145000 (3)	HRPT2, C1orf28	607393	1q25-q31	Endocrine
743	Hyperparathyroidism-jaw tumor syndrome, 145001 (3)	HRPT2, C1orf28	607393	1q25-q31	Endocrine
743	Hyperparathyroidism, neonatal, 239200 (3)	CASR, HHC1, PCAR1, FIH	601199	3q13.3-q21	Endocrine
746	Hyperphenylalaninemia due to pterin-4a-carbinolamine dehydratase deficiency, 264070 (3)	PCBD, DCOH	126090	10q22	Metabolic
746	Hyperphenylalaninemia, mild (3)	PAH, PKU1	261600	12q24.1	Metabolic
748	Hyperproinsulinemia, familial (3)	INS	176730	11p15.5	Endocrine
749	Hyperprolinemia, type I, 239500 (3)	PRODH, PRODH2, SCZD4	606810	22q11.2	Metabolic
749	Hyperprolinemia, type II, 239510 (3)	ALDH4A1, ALDH4, P5CDH	606811	1p36	Metabolic
750	Hyperproreninemia (3)	REN	179820	1q32	Cardiovascular
751	Hyperprothrombinemia (3)	F2	176930	11p11-q12	Hematological
752	Hypertension, diastolic, resistance to, 608622 (3)	KCNMB1	603951	5q34	Cardiovascular
752	Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115 (3)	NR3C2, MLR, MCR	600983	4q31.1	Cardiovascular
752	Hypertension, essential, 145500 (3)	AGTR1, AGTR1A, AT2R1	106165	3q21-q25	Cardiovascular
752	Hypertension, essential, 145500 (3)	PTGIS, CYP8A1, PGIS, CYP8	601699	20q13.11-q13.13	Cardiovascular
752	Hypertension, essential, salt-sensitive, 145500 (3)	ADD1	102680	4p16.3	Cardiovascular
752	Hypertension, essential, susceptibility to, 145500 (3)	AGT, SERPINA8	106150	1q42-q43	Cardiovascular
752	Hypertension, essential, susceptibility to, 145500 (3)	ECE1	600423	1p36.1	Cardiovascular
752	Hypertension, essential, susceptibility to, 145500 (3)	GNB3	139130	12p13	Cardiovascular
752	Hypertension, insulin resistance-related, susceptibility to, 125853 (3)	RETN, RSTN, FIZZ3	605565	19p13.2	Cardiovascular

752	Hypertension, mild low-renin (3)	HSD11B2, HSD11K	218030	16q22	Cardiovascular
752	Hypertension, pregnancy-induced, 189800 (3)	NOS3	163729	7q36	Cardiovascular
752	Hypertension, salt-sensitive essential, susceptibility to, 145500 (3)	CYP3A5, P450PCN3	605325	7q22.1	Cardiovascular
752	Hypertension, susceptibility to, 145500 (3)	NOS3	163729	7q36	Cardiovascular
754	Hyperthroidism, congenital (3)	TSHR	603372	14q31	Endocrine
755	Hyperthyroidism, congenital (3)	TPO, TPX	606765	2p25	Endocrine
757	Hypertriglyceridemia, one form (3)	APOA1	107680	11q23	Metabolic
757	Hypertriglyceridemia, susceptibility to, 145750 (3)	APOA5	606368	11q23	Metabolic
757	Hypertriglyceridemia, susceptibility to, 145750 (3)	LIPI, LPDL, PRED5	609252	21q11.2	Metabolic
757	Hypertriglyceridemia, susceptibility to, 145750 (3)	RP1, ORP1	603937	8q11-q13	Metabolic
758	Hypertrypsinemia, neonatal (3)	CFTR, ABCC7, CF, MRP7	602421	7q31.2	Gastrointestinal
759	Hyperuricemic nephropathy, familial juvenile, 162000 (3)	UMOD, HNFJ, FJHN, MCKD2, ADMCKD2	191845	16p12.3	Renal
761	Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 (3)	CYP11B2	124080	8q21	Metabolic
761	Hypoaldosteronism, congenital, due to CMO II deficiency (3)	CYP11B2	124080	8q21	Metabolic
762	Hypoalphalipoproteinemia (3)	APOA1	107680	11q23	Metabolic
763	Hypobetalipoproteinemia (3)	APOB, FLDB	107730	2p24	Metabolic
764	Hypocalcemia, autosomal dominant, 146200 (3)	CASR, HHC1, PCAR1, FIH	601199	3q13.3-q21	Endocrine
764	Hypocalcemia, autosomal dominant, with Bartter syndrome (3)	CASR, HHC1, PCAR1, FIH	601199	3q13.3-q21	Endocrine
765	Hypocalciuric hypercalcemia, type I, 145980 (3)	CASR, HHC1, PCAR1, FIH	601199	3q13.3-q21	Endocrine
766	Hypoceruloplasminemia, hereditary, 604290 (3)	CP	117700	3q23-q24	Metabolic
767	Hypochondroplasia, 146000 (3)	FGFR3, ACH	134934	4p16.3	Skeletal
768	Hypochromic microcytic anemia (3)	HBA2	141850	16pter-p13.3	Hematological
769	Hypodontia, 106600 (3)	PAX9	167416	14q12-q13	Skeletal
769	Hypodontia, autosomal dominant, 106600 (3)	MSX1, HOX7, HYD1, OFC5	142983	4p16.1	Skeletal
769	Hypodontia with orofacial cleft, 106600 (3)	MSX1, HOX7, HYD1, OFC5	142983	4p16.1	Skeletal
770	Hypofibrinogenemia, gamma type (3)	FGG	134850	4q28	Hematological
771	Hypoglobulinemia and absent B cells (3)	BLNK, SLP65	604515	10q23.2	Immunological
772	Hypoglycemia of infancy, leucine-sensitive, 240800 (3)	ABCC8, SUR, PHHI, SUR1	600509	11p15.1	Metabolic
772	Hypoglycemia of infancy, persistent hyperinsulinemic, 256450 (3)	ABCC8, SUR, PHHI, SUR1	600509	11p15.1	Metabolic
773	Hypogonadism, hypergonadotropic (3)	LHB	152780	19q13.32	Endocrine
774	Hypogonadotropic hypogonadism, 146110 (3)	GPR54	604161	19p13.3	Endocrine
774	Hypogonadotropic hypogonadism, 146110 (3)	NELF	608137	9q34.3	Endocrine
774	Hypogonadotropic hypogonadism (3)	GNRHR, LHRHR	138850	4q21.2	Endocrine
774	Hypogonadotropic hypogonadism (3)	LHCGR	152790	2p21	Endocrine
775	Hypohaptoglobinemia (3)	HP	140100	16q22.1	Hematological
776	Hypokalemic periodic paralysis, 170400 (3)	CACNA1S, CACNL1A3, CCHL1A3	114208	1q32	Renal
776	Hypokalemic periodic paralysis, 170400 (3)	KCNE3, HOKPP	604433	11q13-q14	Renal
776	Hypokalemic periodic paralysis, 170400 (3)	SCN4A, HYPP, NAC1A	603967	17q23.1-q25.3	Renal
777	Hypolactasia, adult type, 223100 (3)	LCT, LAC, LPH	603202	2q21	Metabolic
777	Hypolactasia, adult type, 223100 (3)	MCM6	601806	2q21	Metabolic
778	Hypomagnesemia-2, renal, 154020 (3)	FXYD2, ATP1G1, HOMG2	601814	11q23	Renal
778	Hypomagnesemia, primary, 248250 (3)	CLDN16, PCLN1	603959	3q27	Renal
778	Hypomagnesemia with secondary hypocalcemia, 602014 (3)	TRPM6, CHAK2	607009	9q22	Renal
779	Hypoparathyroidism, autosomal dominant(3)	PTH	168450	11p15.3-p15.1	Endocrine
779	Hypoparathyroidism, autosomal recessive (3)	PTH	168450	11p15.3-p15.1	Endocrine
779	Hypoparathyroidism, familial isolated, 146200 (3)	GCMB	603716	6p24.2	Endocrine
780	Hypoparathyroidism-retardation-dysmorphism syndrome, 241410 (3)	TBCE, KCS, KCS1, HRD	604934	1q42-q43	Endocrine
781	Hypoparathyroidism, sensorineural deafness, and renal dysplasia, 146255 (3)	GATA3, HDR	131320	10p15	Endocrine
782	Hypophosphatasia, childhood, 241510 (3)	ALPL, HOPS, TNSALP	171760	1p36.1-p34	Metabolic

782	Hypophosphatasia, infantile, 241500 (3)	ALPL, HOPS, TNSALP	171760	1p36.1-p34	Metabolic
783	Hypophosphatemia, type III (3)	CLCN5, CLCK2, NPHL2, DENTS	300008	Xp11.22	Metabolic
783	Hypophosphatemia, X-linked, 307800 (3)	PHEX, HYP, HPDR1	300550	Xp22.2-p22.1	Metabolic
784	Hypophosphatemic rickets, autosomal dominant, 193100 (3)	FGF23, ADHR, HPDR2, PHPTC	605380	12p13.3	Bone
785	Hypoplastic enamel pitting, localized, 608563 (3)	ENAM	606585	4q21	Connective tissue
2785	Hypoplastic left heart syndrome, 241550 (3)	GJA1, CX43, ODDD, SDTY3, ODOD	121014	6q21-q23.2	Cardiovascular
786	Hypoprothrombinemia (3)	F2	176930	11p11-q12	Hematological
787	Hypothyroidism, autoimmune, 140300 (3)	CTLA4	123890	2q33	Endocrine
787	Hypothyroidism, congenital, 274400 (3)	SLC5A5, NIS	601843	19p13.2-p12	Endocrine
787	Hypothyroidism, congenital, due to DUOX2 deficiency, 607200 (3)	DUOX2, THOX2	606759	15q15.3	Endocrine
787	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700 (3)	PAX8	167415	2q12-q14	Endocrine
787	Hypothyroidism, congenital, due to TSH resistance, 275200 (3)	TSHR	603372	14q31	Endocrine
787	Hypothyroidism, hereditary congenital (3)	TG, AITD3	188450	8q24.2-q24.3	Endocrine
787	Hypothyroidism, nongoitrous (3)	TSHB	188540	1p13	Endocrine
787	Hypothyroidism, subclinical (3)	TSHR	603372	14q31	Endocrine
788	Hypotrichosis, congential, with juvenile macular dystrophy, 601553 (3)	CDH3, CDHP, PCAD, HJMD	114021	16q22.1	Dermatological
788	Hypotrichosis, localized, autosomal recessive, 607903 (3)	DSG4, LAH	607892	18q12	Dermatological
788	Hypotrichosis-lymphedema-telangiectasia syndrome, 607823 (3)	SOX18, HLTS	601618	20q13.33	Dermatological
788	Hypotrichosis simplex of scalp, 146520 (3)	CDSN, HTSS	602593	6p21.3	Dermatological
791	Hypouricemia, renal, 220150 (3)	SLC22A12, OAT4L, URAT1	607096	11q13	Renal
792	Hystrix-like ichthyosis with deafness, 602540 (3)	GJB2, CX26, DFNB1, PPK, DFNA3, KID, HID	121011	13q11-q12	multiple
793	Ichthyosiform erythroderma, congenital, 242100 (3)	TGM1, ICR2, LI1	190195	14q11.2	Dermatological
793	Ichthyosiform erythroderma, congenital, nonbullous, 1, 242100 (3)	ALOX12B	603741	17pter-p13.1	Dermatological
793	Ichthyosiform erythroderma, congenital, nonbullous, 1, 242100 (3)	ALOXE3	607206	17p13.1	Dermatological
794	Ichthyosis bullosa of Siemens, 146800 (3)	KRT2A, KRT2E	600194	12q11-q13	Dermatological
794	Ichthyosis, congenital, autosomal recessive (3)	ICHYN	609383	5q33	Dermatological
794	Ichthyosis, cyclic, with epidermolytic hyperkeratosis, 607602 (3)	KRT10	148080	17q21-q22	Dermatological
794	Ichthyosis, harlequin, 242500 (3)	ABCA12, ICR2B, LI2	607800	2q34	Dermatological
794	Ichthyosis histrix, Curth-Macklin type, 146590 (3)	KRT1	139350	12q13	Dermatological
794	Ichthyosis, lamellar 2, 601277 (3)	ABCA12, ICR2B, LI2	607800	2q34	Dermatological
794	Ichthyosis, lamellar, autosomal recessive, 242300 (3)	TGM1, ICR2, LI1	190195	14q11.2	Dermatological
794	Ichthyosis, X-linked (3)	STS, ARSC1, ARSC, SSDD	308100	Xp22.32	Dermatological
795	ICOS deficiency, 607594 (3)	ICOS, AILIM	604558	2q33	Immunological
797	IgE levels QTL, 147050 (3)	PHF11, NYREN34	607796	13q14.1	Immunological
798	IgG2 deficiency, selective (3)	IGHG2	147110	14q32.33	Immunological
799	IgG receptor I, phagocytic, familial deficiency of (3)	FCGR1A, IGFR1, CD64	146760	1q21.2-q21.3	Immunological
801	Immunodeficiency-centromeric instability-facial anomalies syndrome, 242860 (3)	DNMT3B, ICF	602900	20q11.2	multiple
802	Immunodeficiency due to defect in CD3-epsilon (3)	CD3E	186830	11q23	Immunological
802	Immunodeficiency due to defect in CD3-gamma (3)	CD3G	186740	11q23	Immunological
802	Immunodeficiency with hyper-IgM, type 2, 605258 (3)	AICDA, AID, HIGM2	605257	12p13	Immunological
802	Immunodeficiency with hyper-IgM, type 3, 606843 (3)	TNFRSF5, CD40	109535	20q12-q13.2	Immunological
802	Immunodeficiency with hyper IgM, type 4, 608106 (3)	UNG, DGU, HIGM4	191525	12q23-q24.1	Immunological
802	Immunodeficiency, X-linked, with hyper-IgM, 308230 (3)	TNFSF5, CD40LG, HIGM1, IGM	300386	Xq26	Immunological
803	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 (3)	FOXP3, IPEX, AIID, XPID, PIDX	300292	Xp11.23-q13.3	multiple
804	Immunoglobulin A deficiency, 609529 (3)	TNFRSF14B, TACI	604907	17p11.2	Immunological
805	Inclusion body myopathy-3, 605637 (3)	MYH2	160740	17p13.1	Muscular

805	Inclusion body myopathy, autosomal recessive, 600737 (3)	GNE, GLCNE, IBM2, DMRV, NM	603824	9p12-p11	Muscular
805	Inclusion body myopathy with early-onset Paget disease and frontotemporal dementia, 167320 (3)	VCP, IBMPFD	601023	9p13-p12	Muscular
806	Incontinentia pigmenti, type II, 308300 (3)	IKBKG, NEMO, FIP3, IP2	300248	Xq28	Dermatological
807	Infantile spasm syndrome, 308350 (3)	ARX, ISSX, PRTS, MRXS1, MRX36, MRX54	300382	Xp22.13	Neurological
809	Infundibular hypoplasia and hypopituitarism (3)	SOX3, MRGH	313430	Xq26.3	Endocrine
810	Inosine triphosphatase deficiency (3)	ITPA	147520	20p	Metabolic
811	Insensitivity to pain, congenital, with anhidrosis, 256800 (3)	NTRK1, TRKA, MTC	191315	1q21-q22	Neurological
812	Insomnia (3) ()	GABRB3	137192	15q11.2-q12	Psychiatric
812	Insomnia, fatal familial, 600072 (3)	PRNP, PRIP	176640	20pter-p12	Psychiatric
814	Insulin resistance, severe, digenic, 604367 (3)	PPARG, PPARG1, PPARG2	601487	3p25	Metabolic
814	Insulin resistance, severe, digenic, 604367 (3)	PPP1R3A, PPP1R3	600917	7q11.23-q21.11	Metabolic
814	Insulin resistance, susceptibility to (3)	PTPN1, PTP1B	176885	20q13.1-q13.2	Metabolic
816	Interleukin-2 receptor, alpha chain, deficiency of (3)	IL2RA, IL2R	147730	10p15-p14	Immunological
817	Intervertebral disc disease, susceptibility to, 603932 (3)	COL9A2, EDM2	120260	1p33-p32.2	Neurological
817	Intervertebral disc disease, susceptibility to, 603932 (3)	COL9A3, EDM3, IDD	120270	20q13.3	Neurological
819	Intrauterine and postnatal growth retardation (3)	IGF1R	147370	15q25-q26	Developmental
819	Intrauterine and postnatal growth retardation (3)	IGF2	147470	11p15.5	Developmental
820	Intrinsic factor deficiency, 261000 (3)	GIF, IF	609342	11q13	Hematological
821	IRAK4 deficiency, 607676 (3)	IRAK4, REN64	606883	12q12	Immunological
822	Iridogoniodysgenesis, 601631 (3)	FOXC1, FKHL7, FREAC3	601090	6p25	Ophthamological
822	Iridogoniodysgenesis syndrome-2, 137600 (3)	PITX2, IDG2, RIEG1, RGS, IGDS2	601542	4q25-q26	Ophthamological
823	Iris hypoplasia and glaucoma (3)	FOXC1, FKHL7, FREAC3	601090	6p25	Ophthamological
824	Iron deficiency anemia, susceptibility to (3)	TF	190000	3q21	Hematological
824	Iron overload, autosomal dominant (3)	FTH1, FTHL6	134770	11q12-q13	Hematological
825	Isolated growth hormone deficiency, Illig type with absent GH and Kowarski type with bioinactive GH (3)	GH1, GHN	139250	17q22-q24	Endocrine
826	Isovaleric acidemia, 243500 (3)	IVD	607036	15q14-q15	Metabolic
827	Jackson-Weiss syndrome, 123150 (3)	FGFR1, FLT2, KAL2	136350	8p11.2-p11.1	Skeletal
827	Jackson-Weiss syndrome, 123150 (3)	FGFR2, BEK, CFD1, JWS	176943	10q26	Skeletal
829	Jensen syndrome, 311150 (3)	TIMM8A, DFN1, DDP, MTS, DDP1	300356	Xq22	Neurological
830	Jervell and Lange-Nielsen syndrome, 220400 (3)	KCNE1, JLNS, LQT5	176261	21q22.1-q22.2	multiple
830	Jervell and Lange-Nielsen syndrome, 220400 (3)	KCNQ1, KCNA9, LQT1, KVLQT1, ATFB1	607542	11p15.5	multiple
831	Joubert syndrome, 213300 (3)	NPHP1, NPH1, SLSN1	607100	2q13	multiple
831	Joubert syndrome-3, 608629 (3)	AHI1	608894	6q23.3	multiple
832	Juberg-Marsidi syndrome, 309590 (3)	ATRX, XH2, XNP, MRXS3, SHS	300032	Xq13	multiple
833	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 (3)	MADH4, DPC4, SMAD4, JIP	600993	18q21.1	Cancer
835	Kallikrein, decreased urinary activity of (3)	KLK1, KLKR	147910	19q13.2-q13.4	Renal
836	Kallmann syndrome 2, 147950 (3)	FGFR1, FLT2, KAL2	136350	8p11.2-p11.1	multiple
836	Kallmann syndrome (3)	KAL1, KMS, ADMLX	308700	Xp22.3	multiple
837	Kanzaki disease, 609242 (3)	NAGA	104170	22q11	Metabolic
838	Kaposi sarcoma, susceptibility to, 148000 (3)	IL6, IFNB2, BSF2	147620	7p21	Immunological
839	Kappa light chain deficiency (3)	IGKC	147200	2p12	Immunological
840	Kartagener syndrome, 244400 (3)	DNAH11, DNAHC11	603339	Chr.7	multiple
840	Kartagener syndrome, 244400 (3)	DNAH5, HL1, PCD, CILD3	603335	5p15-p14	multiple
840	Kartagener syndrome, 244400 (3)	DNAI1, CILD1, ICS, PCD	604366	9p21-p13	multiple
841	Kenny-Caffey syndrome-1, 244460 (3)	TBCE, KCS, KCS1, HRD	604934	1q42-q43	multiple

842	Keratitis, 148190 (3)	PAX6, AN2, MGDA	607108	11p13	Ophthamological
843	Keratitis-ichthyosis-deafness syndrome, 148210 (3)	GJB2, CX26, DFNB1, PPK, DFNA3, KID, HID	121011	13q11-q12	multiple
844	Keratoconus, 148300 (3)	VSX1, RINX, PPCD, PPD, KTCN	605020	20p11.2	Ophthamological
845	Keratoderma, palmoplantar, with deafness, 148350 (3)	GJB2, CX26, DFNB1, PPK, DFNA3, KID, HID	121011	13q11-q12	Dermatological
847	Keratosis follicularis spinulosa decalvans, 308800 (3)	SAT, SSAT, KFSD	313020	Xp22.1	Dermatological
847	Keratosis palmoplantaria striata, 148700 (3)	KRT1	139350	12q13	Dermatological
847	Keratosis palmoplantaris striata I, 148700 (3)	DSG1	125670	18q12.1-q12.2	Dermatological
847	Keratosis palmoplantaris striata II (3)	DSP, KPPS2, PPKS2	125647	6p24	Dermatological
847	Keratosis palmoplantaris striata III, 607654 (3)	KRT1	139350	12q13	Dermatological
848	Ketoacidosis due to SCOT deficiency (3)	SCOT, OXCT	245050	5p13	Metabolic
849	Keutel syndrome, 245150 (3)	MGP, NTI	154870	12p13.1-p12.3	multiple
850	Kindler syndrome, 173650 (3)	KIND1, URP1, C20orf42	607900	20p13	Dermatological
851	Kininogen deficiency (3)	KNG	228960	3q27	Hematological
853	Klippel-Trenaunay syndrome, 149000 (3)	VG5Q, HUS84971, FLJ10283	608464	5q13.3	multiple
854	Kniest dysplasia, 156550 (3)	COL2A1	120140	12q13.11-q13.2	Skeletal
855	Knobloch syndrome, 267750 (3)	COL18A1, KNO	120328	21q22.3	multiple
856	Krabbe disease, 245200 (3)	GALC	606890	14q31	Neurological
857	L-2-hydroxyglutaric aciduria, 236792 (3)	L2HGDH, C14orf160	609584	14q22.1	Metabolic
858	Lactate dehydrogenase-B deficiency (3)	LDHB	150100	12p12.2-p12.1	Metabolic
859	Lacticacidemia due to PDX1 deficiency, 245349 (3)	PDX1	608769	11p13	Metabolic
862	Langer mesomelic dysplasia, 249700 (3)	SHOX, GCFX, SS, PHOG	312865	Xpter-p22.32	Skeletal
862	Langer mesomelic dysplasia, 249700 (3)	SHOXY	400020	Ypter-p11.2	Skeletal
863	Laron dwarfism, 262500 (3)	GHR	600946	5p13-p12	Skeletal
865	Larson syndrome, 150250 (3)	FLNB, SCT, AOI	603381	3p14.3	Skeletal
868	Laryngoonychocutaneous syndrome, 245660 (3)	LAMA3, LOCS	600805	18q11.2	multiple
869	Lathosterolosis, 607330 (3)	SC5DL, ERG3	602286	11q23.3	Metabolic
870	LCHAD deficiency (3)	HADHA, MTPA	600890	2p23	Metabolic
871	Lead poisoning, susceptibility to (3)	ALAD	125270	9q34	Metabolic
872	Leanness, inherited (3)	AGRP, ART, AGRT	602311	16q22	Nutritional
873	Leber congenital amaurosis, 204000 (3)	CRB1, RP12	604210	1q31-q32.1	Ophthamological
873	Leber congenital amaurosis, 204000 (3)	CRX, CORD2, CRD	602225	19q13.3	Ophthamological
873	Leber congenital amaurosis, 204000 (3)	RPGRIP1, LCA6, CORD9	605446	14q11	Ophthamological
873	Leber congenital amaurosis-2, 204100 (3)	RPE65, RP20	180069	1p31	Ophthamological
873	Leber congenital amaurosis, 604393 (3)	AIPL1, LCA4	604392	17p13.1	Ophthamological
873	Leber congenital amaurosis, type I, 204000 (3)	GUCY2D, GUC2D, LCA1, CORD6	600179	17p13.1	Ophthamological
873	Leber congenital amaurosis, type III, 604232 (3)	RDH12, LCA3	608830	14q23.3	Ophthamological
874	Left-right axis malformations (3)	ACVR2B	602730	3p22-p21.3	Developmental
874	Left-right axis malformations (3)	EBAF, TGFB4, LEFTY2, LEFTA, LEFTYA	601877	1q42.1	Developmental
875	Left ventricular noncompaction, familial isolated, 1, 604169 (3)	DTNA, D18S892E, DRP3, LVNC1	601239	18q12.1-q12.2	Cardiovascular
875	Left ventricular noncompaction with congenital heart defects, 606617 (3)	DTNA, D18S892E, DRP3, LVNC1	601239	18q12.1-q12.2	Cardiovascular
876	Legionaire disease, susceptibility to, 608556 (3)	TLR5, TIL3	603031	1q41-q42	Immunological
877	Leigh syndrome, 256000 (3)	BCS1L, FLNMS, GRACILE	603647	2q33	Neurological
877	Leigh syndrome, 256000 (3)	DLD, LAD, PHE3	238331	7q31-q32	Neurological
877	Leigh syndrome, 256000 (3)	NDUFS3	603846	11p11.11	Neurological
877	Leigh syndrome, 256000 (3)	NDUFS4, AQDQ	602694	5q11.1	Neurological
877	Leigh syndrome, 256000 (3)	NDUFS7, PSST	601825	19p13	Neurological
877	Leigh syndrome, 256000 (3)	NDUFS8	602141	11q13	Neurological

877	Leigh syndrome, 256000 (3)	NDUFV1, UQOR1	161015	11q13	Neurological
877	Leigh syndrome, 256000 (3)	SDHA, SDH2, SDHF	600857	5p15	Neurological
877	Leigh syndrome, due to COX deficiency, 256000 (3)	SURF1	185620	9q34	Neurological
877	Leigh syndrome due to cytochrome c oxidase deficiency, 256000 (3)	COX15	603646	10q24	Neurological
877	Leigh syndrome, French-Canadian type, 220111 (3)	LRPPRC, LRP130, LSFC	607544	2p21-p16	Neurological
877	Leigh syndrome, X-linked, 308930 (3)	PDHA1, PHE1A	312170	Xp22.2-p22.1	Neurological
878	Leiomyomatosis and renal cell cancer, 605839 (3)	FH	136850	1q42.1	Cancer
878	Leiomyomatosis, diffuse, with Alport syndrome, 308940 (3)	COL4A6	303631	Xq22.3	Cancer
880	Leopard syndrome, 151100 (3)	PTPN11, PTP2C, SHP2, NS1	176876	12q24.1	multiple
881	Leprechaunism, 246200 (3)	INSR	147670	19p13.2	Developmental
882	Leprosy, susceptibility to, 607572 (3)	PRKN, PARK2, PDJ	602544	6q25.2-q27	Immunological
883	Leri-Weill dyschondrosteosis, 127300 (3)	SHOX, GCFX, SS, PHOG	312865	Xpter-p22.32	Skeletal
883	Leri-Weill dyschondrosteosis, 127300 (3)	SHOXY	400020	Ypter-p11.2	Skeletal
884	Lesch-Nyhan syndrome, 300322, (3)	HPRT1, HPRT	308000	Xq26-q27.2	Metabolic
886	Leukemia-1, T-cell acute lymphocytic (3)	TAL1, TCL5, SCL	187040	1p32	Cancer
886	Leukemia-2, T-cell acute lymphoblastic (3)	TAL2	186855	9q31	Cancer
886	Leukemia, acute lymphoblastic (3)	FLT3	136351	13q12	Cancer
886	Leukemia, acute lymphoblastic (3)	NBS1, NBS	602667	8q21	Cancer
886	Leukemia, acute lymphoblastic (3)	ZNFN1A1, IK1, LYF1	603023	7p12	Cancer
886	Leukemia, acute lymphoblastic, susceptibility to (3)	HOXD4, HOX4B	142981	2q31-q32	Cancer
886	Leukemia, acute lymphocytic (3)	BCR, CML, PHL, ALL	151410	22q11.21	Cancer
886	Leukemia, acute myeloblastic (3)	ARNT	126110	1q21	Cancer
886	Leukemia, acute myelogenous (3)	KRAS2, RASK2	190070	12p12.1	Cancer
886	Leukemia, acute myelogenous, 601626 (3)	GMPS	600358	3q24	Cancer
886	Leukemia, acute myeloid, 601626 (3)	AF10	602409	10p12	Cancer
886	Leukemia, acute myeloid, 601626 (3)	ARHGEF12, LARG, KIAA0382	604763	11q23.3	Cancer
886	Leukemia, acute myeloid, 601626 (3)	CALM, CLTH	603025	11q14	Cancer
886	Leukemia, acute myeloid, 601626 (3)	CEBPA, CEBP	116897	19q13.1	Cancer
886	Leukemia, acute myeloid, 601626 (3)	CHIC2, BTL	604332	4q11-q12	Cancer
886	Leukemia, acute myeloid, 601626 (3)	FLT3	136351	13q12	Cancer
886	Leukemia, acute myeloid, 601626 (3)	KIT, PBT	164920	4q12	Cancer
886	Leukemia, acute myeloid, 601626 (3)	LPP	600700	3q28	Cancer
886	Leukemia, acute myeloid, 601626 (3)	NPM1	164040	5q35	Cancer
886	Leukemia, acute myeloid, 601626 (3)	NUP214, D9S46E, CAN, CAIN	114350	9q34.1	Cancer
886	Leukemia, acute myeloid, 601626 (3)	RUNX1, CBFA2, AML1	151385	21q22.3	Cancer
886	Leukemia, acute myeloid, 601626 (3)	WHSC1L1, NSD3	607083	8p12	Cancer
886	Leukemia, acute myeloid, reduced survival in (3)	FLT3	136351	13q12	Cancer
886	Leukemia, acute myelomonocytic (3)	AF1Q	604684	1q21	Cancer
886	Leukemia, acute promyelocytic, NPM/RARA type (3)	NPM1	164040	5q35	Cancer
886	Leukemia, acute promyelocytic, NUMA/RARA type (3)	NUMA1	164009	11q13	Cancer
886	Leukemia, acute promyelocytic, PL2F/RARA type (3)	ZNF145, PLZF	176797	11q23.1	Cancer
886	Leukemia, acute promyelocytic, PML/RARA type (3)	PML, MYL	102578	15q22	Cancer
886	Leukemia, acute promyeloyctic, STAT5B/RARA type (3)	STAT5B	604260	17q11.2	Cancer
886	Leukemia, acute T-cell lymphoblastic (3)	AF10	602409	10p12	Cancer
886	Leukemia, acute T-cell lymphoblastic (3)	CALM, CLTH	603025	11q14	Cancer
886	Leukemia, chronic lymphatic, susceptibility to, 151400 (3)	ARL11, ARLTS1	609351	13q14.3	Cancer
886	Leukemia, chronic lymphatic, susceptibility to, 151400 (3)	P2RX7, P2X7	602566	12q24	Cancer
886	Leukemia, chronic myeloid, 608232 (3)	BCR, CML, PHL, ALL	151410	22q11.21	Cancer
886	Leukemia, juvenile myelomonocytic, 607785 (3)	GRAF	605370	5q31	Cancer
886	Leukemia, juvenile myelomonocytic, 607785 (3)	NF1, VRNF, WSS, NFNS	162200	17q11.2	Cancer

886	Leukemia, juvenile myelomonocytic, 607785 (3)	PTPN11, PTP2C, SHP2, NS1	176876	12q24.1	Cancer
886	Leukemia/lymphoma, B-cell, 2 (3)	BCL2	151430	18q21.3	Cancer
886	Leukemia/lymphoma, chronic B-cell, 151400 (3)	CCND1, PRAD1, BCL1	168461	11q13	Cancer
886	Leukemia/lymphoma, T-cell (3)	TCRA	186880	14q11.2	Cancer
886	Leukemia, megakaryoblastic, of Down syndrome, 190685 (3)	GATA1, GF1, ERYF1, NFE1	305371	Xp11.23	Cancer
886	Leukemia, megakaryoblastic, with or without Down syndrome, 190685 (3)	GATA1, GF1, ERYF1, NFE1	305371	Xp11.23	Cancer
886	Leukemia, Philadelphia chromosome-positive, resistant to imatinib (3)	ABL1	189980	9q34.1	Cancer
886	Leukemia, post-chemotherapy, susceptibility to (3)	NQO1, DIA4, NMOR1	125860	16q22.1	Cancer
886	Leukemia, T-cell acute lymphoblastic (3)	NUP214, D9S46E, CAN, CAIN	114350	9q34.1	Cancer
889	Leukocyte adhesion deficiency, 116920 (3)	ITGB2, CD18, LCAMB, LAD	600065	21q22.3	Immunological
891	Leukoencephalopathy with vanishing white matter, 603896 (3)	EIF2B1, EIF2BA	606686	Chr.12	Neurological
891	Leukoencephalopathy with vanishing white matter, 603896 (3)	EIF2B2	606454	14q24	Neurological
891	Leukoencephalopathy with vanishing white matter, 603896 (3)	EIF2B3	606273	1p34.1	Neurological
891	Leukoencephalopathy with vanishing white matter, 603896 (3)	EIF2B5, LVWM, CACH, CLE	603945	3q27	Neurological
891	Leukoencephaly with vanishing white matter, 603896 (3)	EIF2B4	606687	2p23.3	Neurological
894	Leydig cell adenoma, with precocious puberty (3)	LHCGR	152790	2p21	Cancer
895	Lhermitte-Duclos syndrome (3)	PTEN, MMAC1	601728	10q23.31	Cancer
896	Liddle syndrome, 177200 (3)	SCNN1B	600760	16p13-p12	Renal
896	Liddle syndrome, 177200 (3)	SCNN1G, PHA1	600761	16p13-p12	Renal
897	Li Fraumeni syndrome, 151623 (3)	CDKN2A, MTS1, P16, MLM, CMM2	600160	9p21	Cancer
898	Li-Fraumeni syndrome, 151623 (3)	TP53, P53, LFS1	191170	17p13.1	Cancer
898	Li-Fraumeni syndrome, 609265 (3)	CHEK2, RAD53, CHK2, CDS1, LFS2	604373	22q12.1	Cancer
899	LIG4 syndrome, 606593 (3)	LIG4	601837	13q22-q34	multiple
901	Limb-mammary syndrome, 603543 (3)	TP73L, TP63, KET, EEC3, SHFM4, LMS, RHS	603273	3q27	multiple
902	Lipodystrophy, congenital generalized, type 1, 608594 (3)	AGPAT2, LPAAB, BSCL, BSCL1	603100	9q34.3	Metabolic
902	Lipodystrophy, congenital generalized, type 2, 269700 (3)	BSCL2, SPG17	606158	11q13	Metabolic
902	Lipodystrophy, familial partial, 151660 (3)	LMNA, LMN1, EMD2, FPLD, CMD1A, HGPS, LGMD1B	150330	1q21.2	Metabolic
902	Lipodystrophy, familial partial, 151660 (3)	PPARG, PPARG1, PPARG2	601487	3p25	Metabolic
902	Lipodystrophy, familial partial, with decreased subcutaneous fat of face and neck (3)	PPARGC1A, PPARGC1	604517	4p15.1	Metabolic
903	Lipoid adrenal hyperplasia, 201710 (3)	STAR	600617	8p11.2	Endocrine
903	Lipoid congenital adrenal hyperplasia, 201710 (3)	CYP11A, P450SCC	118485	15q23-q24	Endocrine
2903	Lipoid proteinosis, 247100 (3)	ECM1	602201	1q21	Metabolic
904	Lipoma (3)	HMGA2, HMGIC, BABL, LIPO	600698	12q14.3	Cancer
904	Lipoma (3)	LPP	600700	3q28	Cancer
904	Lipoma, sporadic (3)	MEN1	131100	11q13	Cancer
904	Lipomatosis, mutiple, 151900 (3)	HMGA2, HMGIC, BABL, LIPO	600698	12q14.3	Cancer
906	Lipoprotein lipase deficiency (3)	LPL, LIPD	238600	8p22	Metabolic
908	Lissencephaly-1, 607432 (3)	PAFAH1B1, LIS1	601545	17p13.3	Neurological
908	Lissencephaly syndrome, Norman-Roberts type, 257320 (3)	RELN, RL	600514	7q22	Neurological
908	Lissencephaly, X-linked, 300067 (3)	DCX, DBCN, LISX	300121	Xq22.3-q23	Neurological
908	Lissencephaly, X-linked with ambiguous genitalia, 300215 (3)	ARX, ISSX, PRTS, MRXS1, MRX36, MRX54	300382	Xp22.13	Neurological
909	Listeria monocytogenes, susceptibility to (3)	CDH1, UVO	192090	16q22.1	Immunological
910	Loeys-Dietz syndrome, 609192 (3)	TGFBR1	190181	9q33-q34	Connective tissue
910	Loeys-Dietz syndrome, 609192 (3)	TGFBR2, HNPCC6	190182	3p22	Connective tissue
911	Longevity, exceptional, 152430 (3)	CETP	118470	16q21	Developmental
911	Longevity, reduced, 152430 (3)	AKAP10	604694	17p11.1	Developmental

912	Long QT syndrome-1, 192500 (3)	KCNQ1, KCNA9, LQT1, KVLQT1, ATFB1	607542	11p15.5	Cardiovascular
912	Long QT syndrome-2 (3)	KCNH2, LQT2, HERG	152427	7q35-q36	Cardiovascular
912	Long QT syndrome-3, 603830 (3)	SCN5A, LQT3, IVF, HB1, SSS1	600163	3p21	Cardiovascular
912	Long QT syndrome 4, 600919 (3)	ANK2, LQT4	106410	4q25-q27	Cardiovascular
912	Long QT syndrome-5 (3)	KCNE1, JLNS, LQT5	176261	21q22.1-q22.2	Cardiovascular
912	Long QT syndrome-6 (3)	KCNE2, MIRP1, LQT6	603796	21q22.1	Cardiovascular
912	Long QT syndrome-7, 170390 (3)	KCNJ2, HHIRK1, KIR2.1, IRK1, LQT7	600681	17q23.1-q24.2	Cardiovascular
913	Lower motor neuron disease, progressive, without sensory symptoms, 607641 (3)	DCTN1	601143	2p13	Neurological
914	Lowe syndrome, 309000 (3)	OCRL, LOCR, OCRL1, NPHL2	300535	Xq26.1	Metabolic
915	Low renin hypertension, susceptibility to (3)	CYP11B2	124080	8q21	Cardiovascular
916	LPA deficiency, congenital (3)	LPA	152200	6q27	Metabolic
917	Lumbar disc disease, susceptibility to, 603932 (3)	CILP	603489	15q22	Skeletal
918	Lung cancer, 211980 (3)	KRAS2, RASK2	190070	12p12.1	Cancer
918	Lung cancer, 211980 (3)	PPP2R1B	603113	11q22-q24	Cancer
918	Lung cancer, 211980 (3)	SLC22A1L, BWSCR1A, IMPT1	602631	11p15.5	Cancer
918	Lung cancer, somatic, 211980 (3)	MAP3K8, COT, EST, TPL2	191195	10p11.2	Cancer
919	Lupus nephritis, susceptibility to (3)	FCGR2A, IGFR2, CD32	146790	1q21-q23	Connective tissue
920	Lymphangioleiomyomatosis, 606690 (3)	TSC1, LAM	605284	9q34	Respiratory
920	Lymphangioleiomyomatosis, somatic, 606690 (3)	TSC2, LAM	191092	16p13.3	Respiratory
921	Lymphedema and ptosis, 153000 (3)	FOXC2, FKHL14, MFH1	602402	16q24.3	Hematological
921	Lymphedema-distichiasis syndrome, 153400 (3)	FOXC2, FKHL14, MFH1	602402	16q24.3	Hematological
921	Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus (3)	FOXC2, FKHL14, MFH1	602402	16q24.3	Hematological
921	Lymphedema, hereditary I, 153100 (3)	FLT4, VEGFR3, PCL	136352	5q35.3	Hematological
921	Lymphedema, hereditary II, 153200 (3)	FOXC2, FKHL14, MFH1	602402	16q24.3	Hematological
925	Lymphocytic leukemia, acute T-cell (3)	RAP1GDS1	179502	4q21-q25	Cancer
925	Lymphoma, B-cell non-Hodgkin, somatic (3)	ATM, ATA, AT1	607585	11q22.3	Cancer
925	Lymphoma, diffuse large cell (3)	BCL8	601889	15q11-q13	Cancer
925	Lymphoma, follicular (3)	BCL10	603517	1p22	Cancer
925	Lymphoma, MALT (3)	BCL10	603517	1p22	Cancer
925	Lymphoma, mantle cell (3)	ATM, ATA, AT1	607585	11q22.3	Cancer
925	Lymphoma, non-Hodgkin (3)	RAD54B	604289	8q21.3-q22	Cancer
925	Lymphoma, non-Hodgkin (3)	RAD54L, HR54, HRAD54	603615	1p32	Cancer
925	Lymphoma, progression of (3)	FCGR2B, CD32	604590	1q22	Cancer
925	Lymphoma, somatic (3)	MAD1L1, TXBP181	602686	7p22	Cancer
925	Lymphoma, T-cell (3)	MSH2, COCA1, FCC1, HNPCC1	609309	2p22-p21	Cancer
925	Lymphoproliferative syndrome, X-linked, 308240 (3)	SH2D1A, LYP, IMD5, XLP, XLPD	300490	Xq25	Cancer
930	Lynch cancer family syndrome II, 114400 (3)	MSH2, COCA1, FCC1, HNPCC1	609309	2p22-p21	Cancer
931	Lysinuric protein intolerance, 222700 (3)	SLC7A7, LPI	603593	14q11.2	Metabolic
933	Machado-Joseph disease, 109150 (3)	ATXN3, MJD, SCA3	607047	14q24.3-q31	Neurological
935	Macrocytic anemia, refractory, of 5q- syndrome, 153550 (3)	IRF1, MAR	147575	5q31.1	Hematological
936	Macrothrombocytopenia, 300367 (3)	GATA1, GF1, ERYF1, NFE1	305371	Xp11.23	Hematological
2937	Macular corneal dystrophy, 217800 (3)	CHST6, MCDC1	605294	16q22	Ophthamological
937	Macular degeneration, age-related, 1, 603075 (3)	HF1, CFH, HUS	134370	1q32	Ophthamological
937	Macular degeneration, age-related, 1, 603075 (3)	HMCN1, FBLN6, FIBL6	608548	1q24-q25	Ophthamological
937	Macular degeneration, age-related, 3, 608895 (3)	FBLN5, ARMD3	604580	14q32.1	Ophthamological
937	Macular degeneration, juvenile, 248200 (3)	CNGB3, ACHM3	605080	8q21-q22	Ophthamological
937	Macular degeneration, X-linked atrophic (3)	RPGR, RP3, CRD, RP15, COD1	312610	Xp21.1	Ophthamological

2937	Macular dystrophy (3)	RDS, RP7, PRPH2, PRPH, AVMD, AOFMD	179605	6p21.1-cen	Ophthamological
2937	Macular dystrophy, age-related, 2, 153800 (3)	ABCA4, ABCR, STGD1, FFM, RP19	601691	1p21-p13	Ophthamological
2937	Macular dystrophy, autosomal dominant, chromosome 6-linked, 600110 (3)	ELOVL4, ADMD, STGD2, STGD3	605512	6q14	Ophthamological
2937	Macular dystrophy, vitelliform, 608161 (3)	RDS, RP7, PRPH2, PRPH, AVMD, AOFMD	179605	6p21.1-cen	Ophthamological
2937	Macular dystrophy, vitelliform type, 153700 (3)	VMD2	607854	11q13	Ophthamological
938	Maculopathy, bull's-eye, 153870 (3)	VMD2	607854	11q13	Ophthamological
939	Major depressive disorder and accelerated response to antidepressant drug treatment, 608616 (3)	FKBP5, FKBP51	602623	6p21.3-p21.2	Psychiatric
940	Malaria, cerebral, reduced risk of, 248310 (3)	CD36	173510	7q11.2	Immunological
940	Malaria, cerebral, susceptibility to, 248310 (3)	CD36	173510	7q11.2	Immunological
940	Malaria, cerebral, susceptibility to (3)	ICAM1	147840	19p13.3-p13.2	Immunological
940	Malaria, cerebral, susceptibility to (3)	TNF, TNFA	191160	6p21.3	Immunological
940	Malaria, resistance to, 248310 (3)	GYPC, GE, GPC	110750	2q14-q21	Immunological
940	Malaria, resistance to, 248310 (3)	NOS2A, NOS2	163730	17cen-q11.2	Immunological
942	Malignant hyperthermia susceptibility 1, 145600 (3)	RYR1, MHS, CCO	180901	19q13.1	Neurological
942	Malignant hyperthermia susceptibility 5, 601887 (3)	CACNA1S, CACNL1A3, CCHL1A3	114208	1q32	Neurological
943	Malonyl-CoA decarboxylase deficiency, 248360 (3)	MLYCD, MCD	606761	16q24	Metabolic
944	MALT lymphoma (3)	MALT1, MLT	604860	18q21	Cancer
945	Mandibuloacral dysplasia with type B lipodystrophy, 608612 (3)	ZMPSTE24, FACE1, STE24, MADB	606480	1p34	multiple
947	Mannosidosis, alpha-, types I and II, 248500 (3)	MAN2B1, MANB	609458	19cen-q12	Metabolic
947	Mannosidosis, beta, 248510 (3)	MANBA, MANB1	609489	4q22-q25	Metabolic
948	Maple syrup urine disease, type Ia, 248600 (3)	BCKDHA, MSUD1	608348	19q13.1-q13.2	Metabolic
948	Maple syrup urine disease, type Ib (3)	BCKDHB, E1B	248611	6p22-p21	Metabolic
948	Maple syrup urine disease, type II (3)	DBT, BCATE2	248610	1p31	Metabolic
948	Maple syrup urine disease, type III, 248600 (3)	DLD, LAD, PHE3	238331	7q31-q32	Metabolic
950	Marfan syndrome, 154700 (3)	FBN1, MFS1, WMS	134797	15q21.1	Connective tissue
950	Marfan syndrome, atypical (3)	COL1A2	120160	7q22.1	Connective tissue
952	Maroteaux-Lamy syndrome, several forms (3)	ARSB, MPS6	253200	5q11-q13	Metabolic
953	Marshall syndrome, 154780 (3)	COL11A1, STL2	120280	1p21	multiple
955	MASA syndrome, 303350 (3)	L1CAM, CAML1, HSAS1	308840	Xq28	multiple
956	MASP2 deficiency (3)	MASP2	605102	1p36.3-p36.2	Immunological
957	MASS syndrome, 604308 (3)	FBN1, MFS1, WMS	134797	15q21.1	Connective tissue
958	Mast cell leukemia (3)	KIT, PBT	164920	4q12	Cancer
959	Mastocytosis with associated hematologic disorder (3)	KIT, PBT	164920	4q12	Immunological
960	Mast syndrome, 248900 (3)	ACP33, MAST, SPG21	608181	15q21-q22	Neurological
961	May-Hegglin anomaly, 155100 (3)	MYH9, MHA, FTNS, DFNA17	160775	22q11.2	Hematological
962	McArdle disease, 232600 (3)	PYGM	608455	11q13	Metabolic
963	McCune-Albright syndrome, 174800 (3)	GNAS, GNAS1, GPSA, POH, PHP1B, PHP1A, AHO	139320	20q13.2	multiple
964	McKusick-Kaufman syndrome, 236700 (3)	MKKS, HMCS, KMS, MKS, BBS6	604896	20p12	Developmental
965	McLeod syndrome (3)	XK	314850	Xp21.2-p21.1	Hematological
965	McLeod syndrome with neuroacanthosis (3)	XK	314850	Xp21.2-p21.1	Hematological
969	Medullary cystic kidney disease 2, 603860 (3)	UMOD, HNFJ, FJHN, MCKD2, ADMCKD2	191845	16p12.3	Renal
2969	Medullary thyroid carcinoma, 155240 (3)	RET, MEN2A	164761	10q11.2	Cancer
2969	Medullary thyroid carcinoma, familial, 155240 (3)	NTRK1, TRKA, MTC	191315	1q21-q22	Cancer
970	Medulloblastoma, 155255 (3)	PTCH2	603673	1p32	Cancer
970	Medulloblastoma, desmoplastic, 155255 (3)	SUFU, SUFUXL, SUFUH	607035	10q24-q25	Cancer

971	Meesmann corneal dystrophy, 122100 (3)	KRT12	601687	17q12	Ophthamological
971	Meesmann corneal dystrophy, 122100 (3)	KRT3	148043	12q13	Ophthamological
973	Megakaryoblastic leukemia, acute (3)	MKL1, AMKL, MAL	606078	22q13	Cancer
974	Megalencephalic leukoencephalopathy with subcortical cysts, 604004 (3)	MLC1, LVM, VL	605908	22qter	Neurological
975	Megaloblastic anemia-1, Finnish type, 261100 (3)	CUBN, IFCR, MGA1	602997	10p12.1	Hematological
975	Megaloblastic anemia-1, Norwegian type, 261100 (3)	AMN	605799	14q32	Hematological
978	Melanoma (3)	CDK4, CMM3	123829	12q14	Cancer
978	Melanoma and neural system tumor syndrome, 155755 (3)	CDKN2A, MTS1, P16, MLM, CMM2	600160	9p21	Cancer
978	Melanoma, cutaneous malignant, 2, 155601 (3)	CDKN2A, MTS1, P16, MLM, CMM2	600160	9p21	Cancer
978	Melanoma, cutaneous malignant, susceptibility to (3)	XRCC3	600675	14q32.3	Cancer
978	Melanoma, malignant sporadic (3)	STK11, PJS, LKB1	602216	19p13.3	Cancer
978	Melanoma, melignant, somatic (3)	BRAF	164757	7q34	Cancer
979	Meleda disease, 248300 (3)	SLURP1, MDM	606119	8qter	Dermatological
981	Melnick-Needles syndrome, 309350 (3)	FLNA, FLN1, ABPX, NHBP, OPD1, OPD2, FMD, MNS	300017	Xq28	Skeletal
982	Melorheostosis with osteopoikilosis, 155950 (3)	LEMD3, MAN1	607844	12q14	Developmental
984	Memory impairment, susceptibility to (3)	BDNF	113505	11p13	Neurological
985	Meniere disease 156000 (3) ()	COCH, DFNA9	603196	14q12-q13	Neurological
986	Meningioma, 607174 (3)	MN1, MGCR	156100	22q12.3-qter	Cancer
986	Meningioma, 607174 (3)	PTEN, MMAC1	601728	10q23.31	Cancer
986	Meningioma, NF2-related, somatic, 607174 (3)	NF2	607379	22q12.2	Cancer
986	Meningioma, SIS-related (3)	PDGFB, SIS	190040	22q12.3-q13.1	Cancer
987	Meningococcal disease, susceptibility to (3)	MBL2, MBL, MBP1	154545	10q11.2-q21	Immunological
988	Menkes disease, 309400 (3)	ATP7A, MNK, MK, OHS	300011	Xq12-q13	Neurological
990	Mental retardation, nonsyndromic, autosomal recessive, 249500 (3)	PRSS12, BSSP3	606709	4q25-q26	Neurological
990	Mental retardation, nonsyndromic, autosomal recessive, 2A, 607417 (3)	CRBN, MRT2A	609262	3p26.2	Neurological
990	Mental retardation, X-linked, 300425 (3)	NLGN4, KIAA1260, AUTSX2	300427	Xp22.33	Neurological
990	Mental retardation, X-linked, 300458 (3)	MECP2, RTT, PPMX, MRX16, MRX79	300005	Xq28	Neurological
990	Mental retardation, X-linked 30, 300558 (3)	PAK3, MRX30, MRX47	300142	Xq21.3-q24	Neurological
990	Mental retardation, X-linked, 34, 300426 (3)	IL1RAPL, MRX34	300206	Xp22.1-p21.3	Neurological
990	Mental retardation, X-linked 36, 300430 (3)	ARX, ISSX, PRTS, MRXS1, MRX36, MRX54	300382	Xp22.13	Neurological
990	Mental retardation, X-linked (3)	SLC6A8, CRTR	300036	Xq28	Neurological
990	Mental retardation, X-linked-44, 300501 (3)	FTSJ1, JM23, SPB1, MRX44, MRX9	300499	Xp11.23	Neurological
990	Mental retardation, X-linked 45, 300498 (3)	ZNF81, MRX45	314998	Xp22.1-p11	Neurological
990	Mental retardation, X-linked 54, 300419 (3)	ARX, ISSX, PRTS, MRXS1, MRX36, MRX54	300382	Xp22.13	Neurological
990	Mental retardation, X-linked 58, 300218 (3)	TM4SF2, MXS1, A15	300096	Xq11	Neurological
990	Mental retardation, X-linked, 60, 300486 (3)	OPHN1	300127	Xq12	Neurological
990	Mental retardation, X-linked-9, 309549 (3)	FTSJ1, JM23, SPB1, MRX44, MRX9	300499	Xp11.23	Neurological
990	Mental retardation, X-linked, FRAXE type (3)	FMR2, FRAXE, MRX2	309548	Xq28	Neurological
990	Mental retardation, X-linked, JARID1C-related, 300534 (3)	SMCX, MRXJ, DXS1272E, XE169, JARID1C	314690	Xp11.22-p11.21	Neurological
990	Mental retardation, X-linked nonspecific, 309541 (3)	GDI1, RABGD1A, MRX41, MRX48	300104	Xq28	Neurological
990	Mental retardation, X-linked nonspecific, 63, 300387 (3)	FACL4, ACS4, MRX63	300157	Xq22.3	Neurological
990	Mental retardation, X-linked nonspecific, type 19 (3)	RPS6KA3, RSK2, MRX19	300075	Xp22.2-p22.1	Neurological
990	Mental retardation, X-linked nonspecific, type 46, 300436 (3)	ARHGEF6, MRX46, COOL2	300267	Xq26	Neurological
990	Mental retardation, X-linked nonsyndromic (3)	AGTR2	300034	Xq22-q23	Neurological
990	Mental retardation, X-linked nonsyndromic (3)	FGD1, FGDY, AAS	305400	Xp11.21	Neurological
990	Mental retardation, X-linked nonsyndromic (3)	ZNF41	314995	Xp22.1-cen	Neurological

990	Mental retardation, X-linked nonsyndromic, DLG3-related (3)	DLG3, NEDLG, SAP102, MRX	300189	Xq13.1	Neurological
990	Mental retardation, X-linked, Snyder-Robinson type, 309583 (3)	SMS, SRS, MRSR	300105	Xp22.1	Neurological
990	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 (3)	SOX3, MRGH	313430	Xq26.3	Neurological
990	Mental retardation, X-linked, with progressive spasticity, 300279 (3)	MECP2, RTT, PPMX, MRX16, MRX79	300005	Xq28	Neurological
990	Mental retardation, X-linked, with seizures and carrier manifestations, 300397 (3)	SLC6A8, CRTR	300036	Xq28	Neurological
991	Mephenytoin poor metabolizer (3)	CYP2C, CYP2C19	124020	10q24.1-q24.3	Metabolic
992	Merkel cell carcinoma, somatic (3)	SDHD, PGL1	602690	11q23	Cancer
993	Mesangial sclerosis, isolated diffuse, 256370 (3)	WT1	607102	11p13	Renal
995	Mesothelioma (3)	BCL10	603517	1p22	Cancer
996	Metachromatic leukodystrophy, 250100 (3)	ARSA	607574	22q13.31-qter	Neurological
996	Metachromatic leukodystrophy due to deficiency of SAP-1 (3)	PSAP, SAP1	176801	10q22.1	Neurological
997	Metaphyseal chondrodysplasia, Murk Jansen type, 156400 (3)	PTHR1, PTHR	168468	3p22-p21.1	Connective tissue
997	Metaphyseal chondrodysplasia, Schmid type (3)	COL10A1	120110	6q21-q22.3	Connective tissue
997	Metaphyseal dysplasia without hypotrichosis, 250460 (3)	RMRP, RMRPR, CHH	157660	9p21-p12	Connective tissue
999	Methemoglobinemia due to cytochrome b5 deficiency (3)	CYB5	250790	18q23	Hematological
999	Methemoglobinemias, alpha- (3)	HBA1	141800	16pter-p13.3	Hematological
999	Methemoglobinemias, beta- (3)	HBB	141900	11p15.5	Hematological
999	Methemoglobinemia, type I (3)	DIA1	250800	22q13.31-qter	Hematological
999	Methemoglobinemia, type II (3)	DIA1	250800	22q13.31-qter	Hematological
1001	Methionine adenosyltransferase deficiency, autosomal recessive (3)	MAT1A, MATA1, SAMS1	250850	10q22	Metabolic
1002	Methylcobalamin deficiency, cblG type, 250940 (3)	MTR	156570	1q43	Metabolic
1003	Methylmalonate semialdehyde dehydrogenase deficiency (3)	ALDH6A1, MMSDH	603178	14q24.3	Metabolic
1004	Methylmalonic aciduria, mut(0) type, 251000 (3)	MUT, MCM	609058	6p21	Metabolic
1004	Methylmalonic aciduria, vitamin B12-responsive, 251100 (3)	MMAA	607481	4q31.1-q31.2	Metabolic
1004	Methylmalonic aciduria, vitamin B12-responsive, due to defect in synthesis of adenosylcobalamin, cblB complementation type, 251110 (3)	MMAB	607568	12q24	Metabolic
1005	Mevalonicaciduria (3)	MVK, MVLK	251170	12q24	Metabolic
1006	MHC class II deficiency, complementation group B, 209920 (3)	RFXANK	603200	19p12	Immunological
1007	Microcephaly, Amish type, 607196 (3)	SLC25A19, DNC, MUP1, MCPHA	606521	17q25.3	Neurological
1007	Microcephaly, autosomal recessive 1, 251200 (3)	MCPH1	607117	8p23	Neurological
1007	Microcephaly, primary autosomal recessive, 3, 604804 (3)	CDK5RAP2, KIAA1633, MCPH3	608201	9q33.3	Neurological
1007	Microcephaly, primary autosomal recessive, 5, 608716 (3)	ASPM, MCPH5	605481	1q31	Neurological
1007	Microcephaly, primary autosomal recessive, 6, 608393 (3)	CEMPJ, CPAP, MCPH6	609279	13q12.2	Neurological
1009	Microcoria-congenital nephrosis syndrome, 609049 (3)	LAMB2, LAMS	150325	3p21	Ophthamological
1011	Micropenis (3)	LHCGR	152790	2p21	Renal
1012	Microphthalmia, cataracts, and iris abnormalities (3)	CHX10, HOX10	142993	14q24.3	Ophthamological
1012	Microphthalmia, SIX6-related (3)	SIX6	606326	14q23	Ophthamological
1012	Microphthalmia with associated anomalies 2, 300412 (3)	BCOR, KIAA1575, MAA2, ANOP2	300485	Xp11.4	Ophthamological
1013	Migraine, familial hemiplegic, 2, 602481 (3)	ATP1A2, FHM2, MHP2	182340	1q21-q23	Neurological
1013	Migraine, resistance to, 157300 (3)	EDNRA	131243	4q31.2	Neurological
1013	Migraine, susceptibility to, 157300 (3)	ESR1, ESR	133430	6q25.1	Neurological
1013	Migraine without aura, susceptibility to, 157300 (3)	TNF, TNFA	191160	6p21.3	Neurological
1014	Miller-Dieker lissencephaly, 247200 (3)	YWHAE, MDCR, MDS	605066	17p13.3	multiple
1016	Mitochondrial complex I deficiency, 252010 (3)	NDUFS1	157655	2q33-q34	multiple
1016	Mitochondrial complex I deficiency, 252010 (3)	NDUFS2	602985	1q23	multiple
1016	Mitochondrial complex I deficiency, 252010 (3)	NDUFS4, AQDQ	602694	5q11.1	multiple
1016	Mitochondrial complex I deficiency, 252010 (3)	NDUFV1, UQOR1	161015	11q13	multiple
1016	Mitochondrial complex III deficiency, 124000 (3)	BCS1L, FLNMS, GRACILE	603647	2q33	multiple

1016	Mitochondrial complex III deficiency, 124000 (3)	UQCRB, UQBP, QPC	191330	8q22	multiple
3016	Mitochondrial DNA depletion myopathy, 251880 (3)	TK2	188250	16q22	Muscular
5016	Mitochondrial DNA depletion syndrome, 251880 (3)	SUCLA2	603921	13q12.2-q13	multiple
5016	Mitochondrial DNA-depletion syndrome, hepatocerebral form, 251880 (3)	DGUOK, DGK	601465	2p13	multiple
7016	Mitochondrial myopathy and sideroblastic anemia, 600462 (3)	PUS1, MLASA	608109	12q24.33	multiple
1016	Mitochondrial respiratory chain complex II deficiency, 252011 (3)	SDHA, SDH2, SDHF	600857	5p15	multiple
1018	Miyoshi myopathy, 254130 (3)	DYSF, LGMD2B	603009	2p13.3-p13.1	Muscular
1020	MODY5 with nephron agenesis (3)	TCF2, HNF2	189907	17cen-q21.3	Endocrine
1020	MODY5 with non-diabetic renal disease and Mullerian aplasia (3)	TCF2, HNF2	189907	17cen-q21.3	Endocrine
1020	MODY, one form, 125850 (3)	INS	176730	11p15.5	Endocrine
1020	MODY, type I, 125850 (3)	HNF4A, TCF14, MODY1	600281	20q12-q13.1	Endocrine
1020	MODY, type II, 125851 (3)	GCK	138079	7p15-p13	Endocrine
1020	MODY, type III, 600496 (3)	TCF1, HNF1A, MODY3	142410	12q24.2	Endocrine
1020	MODY, type IV (3)	IPF1	600733	13q12.1	Endocrine
1020	MODY, type V, 604284 (3)	TCF2, HNF2	189907	17cen-q21.3	Endocrine
1022	Mohr-Tranebjaerg syndrome, 304700 (3)	TIMM8A, DFN1, DDP, MTS, DDP1	300356	Xq22	Neurological
1023	Molybdenum cofactor deficiency, type A, 252150 (3)	MOCS1, MOCOD	603707	6p21.3	Metabolic
1023	Molybdenum cofactor deficiency, type B, 252150 (3)	MOCS2, MPTS	603708	5q11	Metabolic
1023	Molybdenum cofactor deficiency, type C, 252150 (3)	GPH, KIAA1385, GEPH	603930	14q24	Metabolic
1024	Monilethrix, 158000 (3)	KRTHB1, HB1	602153	12q13	Dermatological
1024	Monilethrix, 158000 (3)	KRTHB6, HB6	601928	12q13	Dermatological
1026	Morning glory disc anomaly (3)	PAX6, AN2, MGDA	607108	11p13	Skeletal
1028	Mowat-Wilson syndrome, 235730 (3)	ZFHX1B, SMADIP1, SIP1	605802	2q22	Developmental
1029	Moyamoya disease 3 (3)	MYMY3	608796	8q24	Neurological
1030	Muckle-Wells syndrome, 191900 (3)	CIAS1, C1orf7, FCU, FCAS	606416	1q44	multiple
1031	Mucoepidermoid salivary gland carcinoma (3)	MAML2, MAM3	607537	11q21	Cancer
1031	Mucoepidermoid salivary gland carcinoma (3)	MECT1, KIAA0616	607536	19p13	Cancer
1032	Mucolipidosis IIIA, 252600 (3)	GNPTAB, GNPTA	607840	4q21-q23	Metabolic
1032	Mucolipidosis IIIC, 252605 (3)	GNPTAG	607838	16p	Metabolic
1032	Mucolipidosis IV, 252650 (3)	MCOLN1, ML4	605248	19p13.3-p13.2	Metabolic
1033	Mucopolysaccharidosis Ih, 607014 (3)	IDUA, IDA	252800	4p16.3	Metabolic
1033	Mucopolysaccharidosis Ih/s, 607015 (3)	IDUA, IDA	252800	4p16.3	Metabolic
1033	Mucopolysaccharidosis II (3)	IDS, MPS2, SIDS	309900	Xq28	Metabolic
1033	Mucopolysaccharidosis Is, 607016 (3)	IDUA, IDA	252800	4p16.3	Metabolic
1033	Mucopolysaccharidosis IVA (3)	GALNS, MPS4A	253000	16q24.3	Metabolic
1033	Mucopolysaccharidosis IVB (3)	GLB1	230500	3p21.33	Metabolic
1033	Mucopolysaccharidosis type IIID, 252940 (3)	GNS, G6S	607664	12q14	Metabolic
1033	Mucopolysaccharidosis type IX, 601492 (3)	HYAL1	607071	3p21.3-p21.2	Metabolic
1033	Mucopolysaccharidosis VII (3)	GUSB, MPS7	253220	7q21.11	Metabolic
1034	Muenke syndrome, 602849 (3)	FGFR3, ACH	134934	4p16.3	Skeletal
1035	Muir-Torre syndrome, 158320 (3)	MLH1, COCA2, HNPCC2	120436	3p21.3	Cancer
1035	Muir-Torre syndrome, 158320 (3)	MSH2, COCA1, FCC1, HNPCC1	609309	2p22-p21	Cancer
1036	Mulibrey nanism, 253250 (3)	TRIM37, MUL, KIAA0898	605073	17q22-q23	multiple
3037	Multiple cutaneous and uterine leiomyomata, 150800 (3)	FH	136850	1q42.1	Cancer
1037	Multiple endocrine neoplasia I (3)	MEN1	131100	11q13	Cancer
1037	Multiple endocrine neoplasia IIA, 171400 (3)	RET, MEN2A	164761	10q11.2	Cancer
1037	Multiple endocrine neoplasia IIB, 162300 (3)	RET, MEN2A	164761	10q11.2	Cancer
5037	Multiple malignancy syndrome (3)	TP53, P53, LFS1	191170	17p13.1	Cancer
9037	Multiple myeloma (3)	IRF4, LSIRF	601900	6p25-p23	Cancer
9037	Multiple myeloma, resistance to, 254500 (3)	LIG4	601837	13q22-q34	Cancer

9038	Multiple sclerosis, susceptibility to, 126200 (3)	MHC2TA, C2TA	600005	16p13	Neurological
9038	Multiple sclerosis, susceptibility to, 126200 (3)	PTPRC, CD45, LCA	151460	1q31-q32	Neurological
9039	Multiple sulfatase deficiency, 272200 (3)	SUMF1, FGE	607939	3p26	Metabolic
1038	Muscle-eye-brain disease, 253280 (3)	POMGNT1, MEB	606822	1p34-p33	multiple
1039	Muscle glycogenosis (3)	PHKA1	311870	Xq13	Muscular
1039	Muscle hypertrophy (3)	GDF8, MSTN	601788	2q32.2	Muscular
1040	Muscular dystrophy, congenital, 1C (3)	FKRP, MDC1C, LGMD2I	606596	19q13.3	Muscular
1040	Muscular dystrophy, congenital, due to partial LAMA2 deficiency, 607855 (3)	LAMA2, LAMM	156225	6q22-q23	Muscular
1040	Muscular dystrophy, congenital merosin-deficient, 607855 (3)	LAMA2, LAMM	156225	6q22-q23	Muscular
1040	Muscular dystrophy, congenital, type 1D, 608840 (3)	LARGE, KIAA0609, MDC1D	603590	22q12.3-q13.1	Muscular
1040	Muscular dystrophy, Fukuyama congenital, 253800 (3)	FCMD	607440	9q31	Muscular
1040	Muscular dystrophy, limb-girdle, type 1A, 159000 (3)	TTID, MYOT	604103	5q31	Muscular
1040	Muscular dystrophy, limb-girdle, type 2A, 253600 (3)	CAPN3, CANP3	114240	15q15.1-q21.1	Muscular
1040	Muscular dystrophy, limb-girdle, type 2B, 253601 (3)	DYSF, LGMD2B	603009	2p13.3-p13.1	Muscular
1040	Muscular dystrophy, limb-girdle, type 2C, 253700 (3)	SGCG, LGMD2C, DMDA1, SCG3	608896	13q12	Muscular
1040	Muscular dystrophy, limb-girdle, type 2D, 608099 (3)	SGCA, ADL, DAG2, LGMD2D, DMDA2	600119	17q12-q21.33	Muscular
1040	Muscular dystrophy, limb-girdle, type 2E, 604286 (3)	SGCB, LGMD2E	600900	4q12	Muscular
1040	Muscular dystrophy, limb-girdle, type 2F, 601287 (3)	SGCD, SGD, LGMD2F, CMD1L	601411	5q33	Muscular
1040	Muscular dystrophy, limb-girdle, type 2G, 601954 (3)	TCAP, LGMD2G, CMD1N	604488	17q12	Muscular
1040	Muscular dystrophy, limb-girdle, type 2H, 254110 (3)	TRIM32, HT2A, LGMD2H	602290	9q31-q34.1	Muscular
1040	Muscular dystrophy, limb-girdle, type 2I, 607155 (3)	FKRP, MDC1C, LGMD2I	606596	19q13.3	Muscular
1040	Muscular dystrophy, limb-girdle, type 2J, 608807 (3)	TTN, CMD1G, TMD, LGMD2J	188840	2q24.3	Muscular
1040	Muscular dystrophy, limb-girdle, type 2K, 609308 (3)	POMT1	607423	9q34.1	Muscular
1040	Muscular dystrophy, limb-girdle, type IC, 607801 (3)	CAV3, LGMD1C	601253	3p25	Muscular
1040	Muscular dystrophy, rigid spine, 1, 602771 (3)	SEPN1, SELN, RSMD1	606210	1p36-p35	Muscular
1040	Muscular dystrophy with epidermolysis bullosa simplex, 226670 (3)	PLEC1, PLTN, EBS1	601282	8q24	Muscular
1041	Myasthenia, familial infantile, 1, 605809 (3)	CMS1A1, FIM1	605809	17p13	Muscular
1042	Myasthenic syndrome (3)	SCN4A, HYPP, NAC1A	603967	17q23.1-q25.3	Muscular
1042	Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931 (3)	CHRNB1, ACHRB, SCCMS, CMS2A, CMS1D	100710	17p12-p11	Muscular
1042	Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931 (3)	CHRNE, SCCMS, CMS2A, FCCMS, CMS1E, CMS1D	100725	17p13-p12	Muscular
1042	Myasthenic syndrome, congenital, associated with acetylcholine receptor deficiency, 608931 (3)	RAPSN, CMS1D, CMS1E	601592	11p11.2-p11.1	Muscular
1042	Myasthenic syndrome, congenital, associated with episodic apnea, 254210 (3)	CHAT, CMS1A2	118490	10q11.2	Muscular
1042	Myasthenic syndrome, congenital, associated with facial dysmorphism and acetylcholine receptor deficiency, 608931 (3)	RAPSN, CMS1D, CMS1E	601592	11p11.2-p11.1	Muscular
1042	Myasthenic syndrome, fast-channel congenital, 608930 (3)	CHRNA1, ACHRD, CMS2A, SCCMS, FCCMS	100690	2q24-q32	Muscular
1042	Myasthenic syndrome, fast-channel congenital, 608930 (3)	CHRND, ACHRD, SCCMS, CMS2A, FCCMS	100720	2q33-q34	Muscular
1042	Myasthenic syndrome, fast-channel congenital, 608930 (3)	CHRNE, SCCMS, CMS2A, FCCMS, CMS1E, CMS1D	100725	17p13-p12	Muscular
1042	Myasthenic syndrome, slow-channel congenital, 601462 (3)	CHRNA1, ACHRD, CMS2A, SCCMS, FCCMS	100690	2q24-q32	Muscular
1042	Myasthenic syndrome, slow-channel congenital, 601462 (3)	CHRNB1, ACHRB, SCCMS, CMS2A, CMS1D	100710	17p12-p11	Muscular
1042	Myasthenic syndrome, slow-channel congenital, 601462 (3)	CHRND, ACHRD, SCCMS, CMS2A, FCCMS	100720	2q33-q34	Muscular

1042	Myasthenic syndrome, slow-channel congenital, 601462 (3)	CHRNE, SCCMS, CMS2A, FCCMS, CMS1E, CMS1D	100725	17p13-p12	Muscular
1043	Mycobacterial and salmonella infections, susceptibility to, 209950 (3)	IL12RB1	601604	19p13.1	Immunological
1043	Mycobacterial infection, atypical, familial disseminated, 209950 (3)	IFNGR1	107470	6q23-q24	Immunological
1043	Mycobacterial infection, atypical, familial disseminated, 209950 (3)	IFNGR2, IFNGT1, IFGR2	147569	21q22.1-q22.2	Immunological
1043	Mycobacterial infection, atypical, familial disseminated, 209950 (3)	STAT1	600555	2q32.2-q32.3	Immunological
1043	Mycobacterium tuberculosis, suceptibility to infection by, 607948 (3)	NRAMP1, NRAMP	600266	2q35	Immunological
1045	Myelodysplasia syndrome-1 (3)	MDS1	600049	3q26	Muscular
1045	Myelodysplastic syndrome (3)	FACL6, ACS2	604443	5q31	Muscular
1045	Myelodysplastic syndrome, preleukemic (3)	IRF1, MAR	147575	5q31.1	Muscular
1046	Myelofibrosis, idiopathic, 254450 (3)	JAK2	147796	9p24	Hematological
1047	Myelogenous leukemia, acute (3)	FACL6, ACS2	604443	5q31	Cancer
1047	Myelogenous leukemia, acute (3)	IRF1, MAR	147575	5q31.1	Cancer
1047	Myeloid leukemia, acute, M4Eo subtype (3)	CBFB	121360	16q22	Cancer
1047	Myeloid malignancy, predisposition to (3)	CSF1R, FMS	164770	5q33.2-q33.3	Cancer
1049	Myelokathexis, isolated (3)	CXCR4, D2S201E, NPY3R, WHIM	162643	2q21	Immunological
1050	Myelomonocytic leukemia, chronic (3)	PDGFRB, PDGFR	173410	5q31-q32	Cancer
1051	Myeloperoxidase deficiency, 254600 (3)	MPO	606989	17q23.1	Immunological
1052	Myeloproliferative disorder with eosinophilia, 131440 (3)	PDGFRB, PDGFR	173410	5q31-q32	Cancer
1053	Myoadenylate deaminase deficiency (3)	AMPD1	102770	1p21-p13	Muscular
1054	Myocardial infarction, decreased susceptibility to (3)	F7	227500	13q34	Cardiovascular
1054	Myocardial infarction susceptibility (3)	APOE, AD2	107741	19q13.2	Cardiovascular
1054	Myocardial infarction, susceptibility to (3)	ACE, DCP1, ACE1	106180	17q23	Cardiovascular
1054	Myocardial infarction, susceptibility to (3)	ALOX5AP, FLAP	603700	13q12	Cardiovascular
1054	Myocardial infarction, susceptibility to (3)	LGALS2	150571	22q13.1	Cardiovascular
1054	Myocardial infarction, susceptibility to (3)	LTA, TNFB	153440	6p21.3	Cardiovascular
1054	Myocardial infarction, susceptibility to (3)	OLR1, LOX1	602601	12p13-p12	Cardiovascular
1054	Myocardial infarction, susceptibility to (3)	THBD, THRM	188040	20p11.2	Cardiovascular
1054	Myocardial infarction, susceptibility to, 608446 (3)	GCLM, GLCLR	601176	1p22.1	Cardiovascular
1054	Myocardial infarction, susceptibility to, 608446 (3)	TNFSF4, GP34, OX4OL	603594	1q25	Cardiovascular
1055	Myoclonic epilepsy, juvenile, 1, 254770 (3)	EFHC1, FLJ10466, EJM1	608815	6p12-p11	Neurological
1055	Myoclonic epilepsy, severe, of infancy, 607208 (3)	GABRG2, GEFSP3, CAE2, ECA2	137164	5q31.1-q33.1	Neurological
1055	Myoclonic epilepsy with mental retardation and spasticity, 300432 (3)	ARX, ISSX, PRTS, MRXS1, MRX36, MRX54	300382	Xp22.13	Neurological
1056	Myoglobinuria/hemolysis due to PGK deficiency (3)	PGK1, PGKA	311800	Xq13	Metabolic
1057	Myokymia with neonatal epilepsy, 606437 (3)	KCNQ2, EBN1	602235	20q13.3	Neurological
1058	Myoneurogastrointestinal encephalomyopathy syndrome, 603041 (3)	ECGF1	131222	22q13.32-qter	multiple
1059	Myopathy, actin, congenital, with cores (3)	ACTA1, ASMA, NEM3, NEM1	102610	1q42.1	Muscular
1059	Myopathy, actin, congenital, with excess of thin myofilaments, 161800 (3)	ACTA1, ASMA, NEM3, NEM1	102610	1q42.1	Muscular
1059	Myopathy, cardioskeletal, desmin-related, with cataract, 608810 (3)	CRYAB, CRYA2, CTPP2	123590	11q22.3-q23.1	Muscular
1059	Myopathy, centronuclear, 160150 (3)	MYF6	159991	12q21	Muscular
1059	Myopathy, congenital (3)	ITGA7	600536	12q13	Muscular
1059	Myopathy, desmin-related, cardioskeletal, 601419 (3)	DES, CMD1I	125660	2q35	Muscular
1059	Myopathy, distal, with anterior tibial onset, 606768 (3)	DYSF, LGMD2B	603009	2p13.3-p13.1	Muscular
1059	Myopathy, distal, with decreased caveolin 3 (3)	CAV3, LGMD1C	601253	3p25	Muscular
1059	Myopathy due to CPT II deficiency, 255110 (3)	CPT2	600650	1p32	Muscular
1059	Myopathy due to phosphoglycerate mutase deficiency (3)	PGAM2, PGAMM	261670	7p13-p12.3	Muscular
1059	Myopathy, Laing distal, 160500 (3)	MYH7, CMH1, MPD1	160760	14q12	Muscular
1059	Myopathy, myosin storage, 608358 (3)	MYH7, CMH1, MPD1	160760	14q12	Muscular
1059	Myopathy, nemaline, 3, 161800 (3)	ACTA1, ASMA, NEM3, NEM1	102610	1q42.1	Muscular

1061	Myotilinopathy, 609200 (3)	TTID, MYOT	604103	5q31	Muscular
1062	Myotonia congenita, atypical, acetazolamide-responsive, 608390 (3)	SCN4A, HYPP, NAC1A	603967	17q23.1-q25.3	Muscular
1062	Myotonia congenita, dominant, 160800 (3)	CLCN1	118425	7q35	Muscular
1062	Myotonia congenita, recessive, 255700 (3)	CLCN1	118425	7q35	Muscular
1062	Myotonia levior, recessive (3)	CLCN1	118425	7q35	Muscular
1063	Myotonic dystrophy, 160900 (3)	DMPK, DM, DMK	605377	19q13.2-q13.3	Muscular
1063	Myotonic dystrophy, type 2, 602668 (3)	ZNF9, CNBP1, DM2, PROMM	116955	3q13.3-q24	Muscular
1064	Myotubular myopathy, X-linked, 310400 (3)	MTM1, MTMX	300415	Xq28	Muscular
1065	Myxoid liposarcoma (3)	DDIT3, GADD153, CHOP10	126337	12q13.1-q13.2	Cancer
1066	Myxoma, intracardiac, 255960 (3)	PRKAR1A, TSE1, CNC1, CAR	188830	17q23-q24	Cardiovascular
1067	N-acetylglutamate synthase deficiency, 237310 (3)	NAGS	608300	17q21.31	Metabolic
1070	Nail-patella syndrome, 161200 (3)	LMX1B, NPS1	602575	9q34.1	multiple
1070	Nail-patella syndrome with open-angle glaucoma, 137750 (3)	LMX1B, NPS1	602575	9q34.1	multiple
1071	Nance-Horan syndrome, 302350 (3)	NHS	300457	Xp22.13	Ophthamological
1073	Narcolepsy, 161400 (3)	HCRT, OX	602358	17q21	Psychiatric
1074	Nasopharyngeal carcinoma, 161550 (3)	TP53, P53, LFS1	191170	17p13.1	Cancer
1075	Nasu-Hakola disease, 221770 (3)	TREM2	605086	6p21.2	Bone
1075	Nasu-Hakola disease, 221770 (3)	TYROBP, PLOSL, DAP12	604142	19q13.1	Bone
1077	Naxos disease, 601214 (3)	JUP, DP3, PDGB	173325	17q21	multiple
1078	Nemaline myopathy, 161800 (3)	TPM2, TMSB, AMCD1, DA1	190990	9p13.2-p13.1	Muscular
1078	Nemaline myopathy 1, autosomal dominant, 161800 (3)	TPM3, NEM1	191030	1q22-q23	Muscular
1078	Nemaline myopathy 2, autosomal recessive, 256030 (3)	NEB, NEM2	161650	2q22	Muscular
1078	Nemaline myopathy, Amish type, 605355 (3)	TNNT1, ANM	191041	19q13.4	Muscular
3079	Neonatal ichthyosis-sclerosing cholangitis syndrome, 607626 (3)	CLDN1, SEMP1	603718	3q28-q29	multiple
1080	Nephrogenic syndrome of inappropriate antidiuresis, 300539 (3)	AVPR2, DIR, DI1, ADHR	300538	Xq28	Renal
1081	Nephrolithiasis, type I, 310468 (3)	CLCN5, CLCK2, NPHL2, DENTS	300008	Xp11.22	Renal
1081	Nephrolithiasis, uric acid, susceptibility to, 605990 (3)	ZNF365, UAN	607818	10q21.2	Renal
1082	Nephronophthisis 2, infantile, 602088 (3)	INVS, INV, NPHP2, NPH2	243305	9q31	Renal
1082	Nephronophthisis 4, 606966 (3)	NPHP4, SLSN4	607215	1p36	Renal
1082	Nephronophthisis, adolescent, 604387 (3)	NPHP3, NPH3	608002	3q22	Renal
1082	Nephronophthisis, juvenile, 256100 (3)	NPHP1, NPH1, SLSN1	607100	2q13	Renal
1084	Nephropathy, chronic hypocomplementemic (3)	HF1, CFH, HUS	134370	1q32	Renal
1085	Nephropathy with pretibial epidermolysis bullosa and deafness, 609057 (3)	CD151, PETA3, SFA1	602243	11p15.5	Renal
1087	Nephrosis-1, congenital, Finnish type, 256300 (3)	NPHS1, NPHN	602716	19q13.1	Renal
1087	Nephrotic syndrome, steroid-resistant, 600995 (3)	PDCN, NPHS2, SRN1	604766	1q25-q31	Renal
1088	Netherton syndrome, 256500 (3)	SPINK5, LEKTI	605010	5q32	Dermatological
1090	Neural tube defects, maternal risk of, 601634 (3)	MTHFD, MTHFC	172460	14q24	Developmental
1091	Neuroblastoma, 256700 (3)	NME1, NM23	156490	17q21.3	Cancer
1091	Neuroblastoma, 256700 (3)	PMX2B, NBPHOX, PHOX2B	603851	4p12	Cancer
1092	Neurodegeneration, pantothenate kinase-associated, 234200 (3)	PANK2, NBIA1, PKAN, HARP	606157	20p13-p12.3	Neurological
1093	Neuroectodermal tumors, supratentorial primitive, with cafe-au-lait spots, 608623 (3)	PMS2, PMSL2, HNPCC4	600259	7p22	multiple
1095	Neurofibromatosis, familial spinal, 162210 (3)	NF1, VRNF, WSS, NFNS	162200	17q11.2	Cancer
1096	Neurofibromatosis-Noonan syndrome, 601321 (3)	NF1, VRNF, WSS, NFNS	162200	17q11.2	Cancer
1097	Neurofibromatosis, type 1 (3)	NF1, VRNF, WSS, NFNS	162200	17q11.2	Cancer
1097	Neurofibromatosis, type 2, 101000 (3)	NF2	607379	22q12.2	Cancer
1097	Neurofibromatosis, type I, with leukemia, 162200 (3)	MSH2, COCA1, FCC1, HNPCC1	609309	2p22-p21	Cancer
1098	Neurofibrosarcoma (3)	MXI1	600020	10q25	Cancer
1099	Neuropathy, congenital hypomyelinating, 1, 605253 (3)	EGR2, KROX20	129010	10q21.1-q22.1	Neurological
1099	Neuropathy, congenital hypomyelinating, 605253 (3)	MPZ, CMT1B, CMTDI3, CHM, DSS	159440	1q22	Neurological

1099	Neuropathy, distal hereditary motor, 608634 (3)	HSPB1, HSP27, CMT2F	602195	7q11.23	Neurological
1099	Neuropathy, distal hereditary motor, type II, 158590 (3)	HSPB8, H11, E2IG1, DHMN2	608014	12q24-qter	Neurological
1099	Neuropathy, hereditary sensory and autonomic, type 1, 162400 (3)	SPTLC1, LBC1, SPT1, HSN1, HSAN	605712	9q22.1-q22.3	Neurological
1099	Neuropathy, hereditary sensory and autonomic, type V, 608654 (3)	NGFB, HSAN5	162030	1p13.1	Neurological
1099	Neuropathy, hereditary sensory, type II, 201300 (3)	HSN2	608620	12p13.33	Neurological
1099	Neuropathy, recurrent, with pressure palsies, 162500 (3)	PMP22, CMT1A, CMT1E, DSS	601097	17p11.2	Neurological
1101	Neutropenia, alloimmune neonatal (3)	FCGR3A, CD16, IGFR3	146740	1q23	Hematological
1101	Neutropenia, congenital, 202700 (3)	ELA2	130130	19p13.3	Hematological
1101	Neutropenia, severe congenital, 202700 (3)	GFI1, ZNF163	600871	1p22	Hematological
1101	Neutropenia, severe congenital, X-linked, 300299 (3)	WAS, IMD2, THC	300392	Xp11.23-p11.22	Hematological
1102	Neutrophil immunodeficiency syndrome, 608203 (3)	RAC2	602049	22q12.3-q13.2	Immunological
1103	Nevo syndrome, 601451 (3)	PLOD, PLOD1	153454	1p36.3-p36.2	Connective tissue
1104	Nevus, epidermal, epidermolytic hyperkeratotic type, 600648 (3)	KRT10	148080	17q21-q22	Dermatological
1105	Newfoundland rod-cone dystrophy, 607476 (3)	RLBP1	180090	15q26	Ophthamological
1106	Nicotine addiction, protection from (3)	CYP2A6, CYP2A3, CYP2A, P450C2A	122720	19q13.2	Psychiatric
1106	Nicotine addiction, susceptibility to, 188890 (3)	CHRNA4, ENFL1	118504	20q13.2-q13.3	Psychiatric
1106	Nicotine dependence, susceptibility to, 188890 (3)	GPR51, GABBR2	607340	9q22.1	Psychiatric
1107	Niemann-Pick disease, type A, 257200 (3)	SMPD1, NPD	607608	11p15.4-p15.1	Metabolic
1107	Niemann-Pick disease, type B, 607616 (3)	SMPD1, NPD	607608	11p15.4-p15.1	Metabolic
1107	Niemann-Pick disease, type C1, 257220 (3)	NPC1, NPC	607623	18q11-q12	Metabolic
1107	Niemann-pick disease, type C2, 607625 (3)	NPC2, HE1	601015	14q24.3	Metabolic
1107	Niemann-Pick disease, type D, 257220 (3)	NPC1, NPC	607623	18q11-q12	Metabolic
1110	Night blindness, congenital stationary (3)	GNAT1	139330	3p21	Ophthamological
1110	Night blindness, congenital stationary, type 1, 310500 (3)	CSNB1, NYX	300278	Xp11.4	Ophthamological
1110	Night blindness, congenital stationary, type 3, 163500 (3)	PDE6B, PDEB, CSNB3	180072	4p16.3	Ophthamological
1110	Night blindness, congenital stationary, X-linked, type 2, 300071 (3)	CACNA1F, CSNB2	300110	Xp11.23	Ophthamological
1110	Night blindness, congenital stationery, rhodopsin-related (3)	RHO, RP4, OPN2	180380	3q21-q24	Ophthamological
1111	Nijmegen breakage syndrome, 251260 (3)	NBS1, NBS	602667	8q21	multiple
1112	Nonaka myopathy, 605820 (3)	GNE, GLCNE, IBM2, DMRV, NM	603824	9p12-p11	Muscular
1113	Noncompaction of left ventricular myocardium, isolated, 300183 (3)	TAZ, EFE2, BTHS, CMD3A, LVNCX	300394	Xq28	Cardiovascular
1114	Non-Hodgkin lymphoma, somatic, 605027 (3)	CASP10, MCH4, ALPS2	601762	2q33-q34	Cancer
1115	Nonsmall cell lung cancer (3)	IRF1, MAR	147575	5q31.1	Cancer
1115	Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in, 211980 (3)	EGFR	131550	7p12.3-p12.1	Cancer
1115	Nonsmall cell lung cancer, somatic (3)	BRAF	164757	7q34	Cancer
1116	Noonan syndrome 1, 163950 (3)	PTPN11, PTP2C, SHP2, NS1	176876	12q24.1	Developmental
1117	Norrie disease (3)	NDP, ND	310600	Xp11.4	Neurological
1118	Norum disease, 245900 (3)	LCAT	606967	16q22.1	Metabolic
1119	Norwalk virus infection, resistance to (3)	FUT2, SE	182100	19q13.3	Immunological
1122	Nucleoside phosphorylase deficiency, immunodeficiency due to (3)	NP	164050	14q13.1	Immunological
1126	Obesity, adrenal insufficiency, and red hair (3)	POMC	176830	2p23.3	Nutritional
1126	Obesity, autosomal dominant, 601665 (3)	MC4R	155541	18q22	Nutritional
1126	Obesity, hyperphagia, and developmental delay (3)	AKR1C2, DDH2, DD2, HAKRD	600450	10p15-p14	Nutritional
1126	Obesity, hyperphagia, and developmental delay (3)	NTRK2, TRKB	600456	9q22.1	Nutritional
1126	Obesity, late-onset, 601665 (3)	AGRP, ART, AGRT	602311	16q22	Nutritional
1126	Obesity, mild, early-onset, 601665 (3)	NR0B2, SHP	604630	1p36.1	Nutritional
1126	Obesity, morbid, with hypogonadism (3)	LEP, OB	164160	7q31.3	Nutritional
1126	Obesity, morbid, with hypogonadism (3)	LEPR, OBR	601007	1p31	Nutritional
1126	Obesity, resistance to (3)	PPARG, PPARG1, PPARG2	601487	3p25	Nutritional
1126	Obesity, severe, 601665 (3)	PPARG, PPARG1, PPARG2	601487	3p25	Nutritional
1126	Obesity, severe, 601665 (3)	SIM1	603128	6q16.3-q21	Nutritional

1126	Obesity, severe, and type II diabetes, 601665 (3)	UCP3	602044	11q13	Nutritional
1126	Obesity, severe, due to leptin deficiency (3)	LEP, OB	164160	7q31.3	Nutritional
1126	Obesity, severe, susceptibility to, 601665 (3)	MC3R	155540	20q13.2	Nutritional
1126	Obesity, susceptibility to, 300306 (3)	SLC6A14, OBX	300444	Xq23-q24	Nutritional
1126	Obesity, susceptibility to, 601665 (3)	ADRB2	109690	5q32-q34	Nutritional
1126	Obesity, susceptibility to, 601665 (3)	ADRB3	109691	8p12-p11.2	Nutritional
1126	Obesity, susceptibility to, 601665 (3)	CART	602606	5q13.2	Nutritional
1126	Obesity, susceptibility to, 601665 (3)	ENPP1, PDNP1, NPPS, M6S1, PCA1	173335	6q22-q23	Nutritional
1126	Obesity, susceptibility to, 601665 (3)	GHRL	605353	3p26-p25	Nutritional
1126	Obesity, susceptibility to, 601665 (3)	UCP1	113730	4q31	Nutritional
1126	Obesity, susceptibility to, 601665 (3)	UCP2	601693	11q13	Nutritional
1126	Obestiy with impaired prohormone processing, 600955 (3)	PCSK1, NEC1, PC1, PC3	162150	5q15-q21	Nutritional
1128	Obsessive-compulsive disorder 1, 164230 (3)	SLC6A4, HTT, OCD1	182138	17q11.1-q12	Psychiatric
1128	Obsessive-compulsive disorder, protection against, 164230 (3)	BDNF	113505	11p13	Psychiatric
1128	Obsessive-compulsive disorder, susceptibility to, 164230 (3)	HTR2A	182135	13q14-q21	Psychiatric
1129	Occipital horn syndrome, 304150 (3)	ATP7A, MNK, MK, OHS	300011	Xq12-q13	multiple
1130	Ocular albinism, Nettleship-Falls type (3)	OA1	300500	Xp22.3	Ophthamological
1130	Oculocutaneous albinism, type II, modifier of (3)	MC1R	155555	16q24.3	Ophthamological
1130	Oculocutaneous albinism, type IV, 606574 (3)	MATP, AIM1	606202	5p13.3	Ophthamological
1132	Oculodentodigital dysplasia, 164200 (3)	GJA1, CX43, ODDD, SDTY3, ODOD	121014	6q21-q23.2	Skeletal
1133	Oculofaciocardiodental syndrome, 300166 (3)	BCOR, KIAA1575, MAA2, ANOP2	300485	Xp11.4	multiple
1135	Oculopharyngeal muscular dystorphy, 164300 (3)	PABPN1, PABP2, PAB2	602279	14q11.2-q13	Muscular
1135	Oculopharyngeal muscular dystrophy, autosomal recessive, 257950 (3)	PABPN1, PABP2, PAB2	602279	14q11.2-q13	Muscular
1136	Odontohypophosphatasia, 146300 (3)	ALPL, HOPS, TNSALP	171760	1p36.1-p34	Bone
1137	Oguchi disease-1, 258100 (3)	SAG	181031	2q37.1	Ophthamological
1137	Oguchi disease-2, 258100 (3)	RHOK, RK, GRK1	180381	13q34	Ophthamological
1138	Oligodendroglioma, 137800 (3)	PTEN, MMAC1	601728	10q23.31	Cancer
1139	Oligodontia, 604625 (3)	PAX9	167416	14q12-q13	Skeletal
1140	Oligodontia-colorectal cancer syndrome, 608615 (3)	AXIN2	604025	17q24	Cancer
1141	Omenn syndrome, 603554 (3)	DCLRE1C, ARTEMIS, SCIDA	605988	10p	Immunological
1141	Omenn syndrome, 603554 (3)	RAG1	179615	11p13	Immunological
1141	Omenn syndrome, 603554 (3)	RAG2	179616	11p13	Immunological
1142	Opitz G syndrome, type I, 300000 (3)	MID1, OGS1, BBBG1, FXY, OSX	300552	Xp22	multiple
1143	Opremazole poor metabolizer (3)	CYP2C, CYP2C19	124020	10q24.1-q24.3	Metabolic
1144	Optic atrophy 1, 165500 (3)	OPA1, NTG, NPG	605290	3q28-q29	Ophthamological
1144	Optic atrophy and cataract, 165300 (3)	OPA3, MGA3	606580	19q13.2-q13.3	Ophthamological
3144	Optic nerve coloboma with renal disease, 120330 (3)	PAX2	167409	10q24.3-q25.1	multiple
5144	Optic nerve hypoplasia/aplasia, 165550 (3)	PAX6, AN2, MGDA	607108	11p13	Ophthamological
1145	Oral-facial-digital syndrome 1, 311200 (3)	OFD1, CXorf5	300170	Xp22.3-p22.2	Skeletal
1146	Ornithine transcarbamylase deficiency, 311250 (3)	OTC	300461	Xp21.1	Metabolic
1147	Orofacial cleft 6, 608864 (3)	IRF6, VWS, LPS, PIT, PPS, OFC6	607199	1q32-q41	Skeletal
1148	Orolaryngeal cancer, multiple, (3)	CDKN2A, MTS1, P16, MLM, CMM2	600160	9p21	Cancer
1149	Oroticaciduria (3)	UMPS, OPRT	258900	3q13	Metabolic
1150	Orthostatic intolerance, 604715 (3)	SLC6A2, NAT1, NET1	163970	16q12.2	Cardiovascular
1151	OSMED syndrome, 215150 (3)	COL11A2, STL3, DFNA13	120290	6p21.3	Bone
1152	Osseous heteroplasia, progressive, 166350 (3)	GNAS, GNAS1, GPSA, POH, PHP1B, PHP1A, AHO	139320	20q13.2	Bone
1153	Ossification of posterior longitudinal ligament of spine, 602475 (3)	ENPP1, PDNP1, NPPS, M6S1, PCA1	173335	6q22-q23	Connective tissue
1154	Osteoarthritis, hand, susceptibility to, 607850 (3)	MATN3, EDM5, HOA	602109	2p24-p23	Connective tissue
1154	Osteoarthritis of hip, female-specific, susceptibility to, 165720 (3)	FRZB, FRZB1, SRFP3	605083	2q31-q33	Connective tissue

1154	Osteoarthritis, susceptibility to, 165720 (3)	ASPN, PLAP1	608135	9q21.3-q22	Connective tissue
1154	Osteoarthrosis, 165720 (3)	COL2A1	120140	12q13.11-q13.2	Connective tissue
1156	Osteogenesis imperfecta, 3 clinical forms, 166200, 166210, 259420 (3)	COL1A2	120160	7q22.1	Bone
1156	Osteogenesis imperfecta, type I, 166200 (3)	COL1A1	120150	17q21.31-q22	Bone
1156	Osteogenesis imperfecta, type II, 166210 (3)	COL1A1	120150	17q21.31-q22	Bone
1156	Osteogenesis imperfecta, type III, 259420 (3)	COL1A1	120150	17q21.31-q22	Bone
1156	Osteogenesis imperfecta, type IV, 166220 (3)	COL1A1	120150	17q21.31-q22	Bone
1157	Osteolysis, familial expansile, 174810 (3)	TNFRSF11A, RANK, ODFR, OFE	603499	18q22.1	Bone
1157	Osteolysis, idiopathic, Saudi type, 605156 (3)	MMP2, CLG4A, MONA	120360	16q13	Bone
1161	Osteopetrosis, autosomal dominant, type I, 607634 (3)	LRP5, BMND1, LRP7, LR3, OPPG, VBCH2	603506	11q13.4	Bone
1161	Osteopetrosis, autosomal dominant, type II, 166600 (3)	CLCN7, CLC7, OPTA2	602727	16p13	Bone
1161	Osteopetrosis, autosomal recessive, 259700 (3)	OSTM1, GL	607649	6q21	Bone
1161	Osteopetrosis, recessive, 259700 (3)	CLCN7, CLC7, OPTA2	602727	16p13	Bone
1161	Osteopetrosis, recessive, 259700 (3)	TCIRG1, TIRC7, OC116, OPTB1	604592	11q13.4-q13.5	Bone
1162	Osteopoikilosis, 166700 (3)	LEMD3, MAN1	607844	12q14	Bone
1163	Osteoporosis, 166710 (3)	COL1A1	120150	17q21.31-q22	Bone
1163	Osteoporosis, 166710 (3)	LRP5, BMND1, LRP7, LR3, OPPG, VBCH2	603506	11q13.4	Bone
1163	Osteoporosis (3)	CALCA, CALC1	114130	11p15.2-p15.1	Bone
1163	Osteoporosis, hypophosphatemic, (3)	SLC17A2, NPT2	182309	5q35	Bone
1163	Osteoporosis, idiopathic, 166710 (3)	COL1A2	120160	7q22.1	Bone
1163	Osteoporosis, postmenopausal, susceptibility, 166710 (3)	CALCR, CRT	114131	7q21.3	Bone
1164	Osteoporosis-pseudoglioma syndrome, 259770 (3)	LRP5, BMND1, LRP7, LR3, OPPG, VBCH2	603506	11q13.4	Bone
1165	Osteoporosis, susceptibility to, 166710 (3)	RIL	603422	5q31.1	Bone
1166	Osteosarcoma (3)	TP53, P53, LFS1	191170	17p13.1	Cancer
1166	Osteosarcoma, somatic, 259500 (3)	CHEK2, RAD53, CHK2, CDS1, LFS2	604373	22q12.1	Cancer
1168	Otopalatodigital syndrome, type I, 311300 (3)	FLNA, FLN1, ABPX, NHBP, OPD1, OPD2, FMD, MNS	300017	Xq28	multiple
1168	Otopalatodigital syndrome, type II, 304120 (3)	FLNA, FLN1, ABPX, NHBP, OPD1, OPD2, FMD, MNS	300017	Xq28	multiple
1170	Ovarian cancer (3)	BRCA1, PSCP	113705	17q21	Cancer
1170	Ovarian cancer (3)	MSH2, COCA1, FCC1, HNPCC1	609309	2p22-p21	Cancer
1170	Ovarian cancer, 604370 (3)	PIK3CA	171834	3q26.3	Cancer
1170	Ovarian cancer, endometrial type (3)	MSH6, GTBP, HNPCC5	600678	2p16	Cancer
1170	Ovarian cancer, somatic, (3)	ERBB2, NGL, NEU, HER2	164870	17q21.1	Cancer
1170	Ovarian carcinoma (3)	CDH1, UVO	192090	16q22.1	Cancer
1170	Ovarian carcinoma (3)	RRAS2, TC21	600098	11pter-p15.5	Cancer
1170	Ovarian carcinoma, endometrioid type (3)	CTNNB1	116806	3p22-p21.3	Cancer
3171	Ovarian dysgenesis 1, 233300 (3)	FSHR, ODG1	136435	2p21-p16	Endocrine
3171	Ovarian dysgenesis 2, 300510 (3)	BMP15, GDF9B, ODG2	300247	Xp11.2	Endocrine
5170	Ovarian hyperstimulation syndrome, gestational, 608115 (3)	FSHR, ODG1	136435	2p21-p16	Endocrine
7170	Ovarian sex cord tumors (3)	FSHR, ODG1	136435	2p21-p16	Cancer
1171	Ovarioleukodystrophy, 603896 (3)	EIF2B2	606454	14q24	Neurological
1171	Ovarioleukodystrophy, 603896 (3)	EIF2B4	606687	2p23.3	Neurological
1171	Ovarioleukodystrophy, 603896 (3)	EIF2B5, LVWM, CACH, CLE	603945	3q27	Neurological
1172	Pachyonychia congenita, Jackson-Lawler type, 167210 (3)	KRT17, PC2, PCHC1	148069	17q12-q21	Dermatological
1172	Pachyonychia congenita, Jackson-Lawler type, 167210 (3)	KRT6B, PC2	148042	12q13	Dermatological
1172	Pachyonychia congenita, Jadassohn-Lewandowsky type, 167200 (3)	KRT16	148067	17q12-q21	Dermatological

1172	Pachyonychia congenita, Jadassohn-Lewandowsky type, 167200 (3)	KRT6A	148041	12q13	Dermatological
1173	Paget disease, juvenile, 239000 (3)	TNFRSF11B, OPG, OCIF	602643	8q24	Bone
1173	Paget disease of bone, 602080 (3)	SQSTM1, P62, PDB3	601530	5q35	Bone
1173	Paget disease of bone, 602080 (3)	TNFRSF11A, RANK, ODFR, OFE	603499	18q22.1	Bone
1174	Pallidopontonigral degeneration, 168610 (3)	MAPT, MTBT1, DDPAC, MSTD	157140	17q21.1	Neurological
1175	Pallister-Hall syndrome, 146510 (3)	GLI3, PAPA, PAPB, ACLS	165240	7p13	multiple
1176	Palmoplantar keratoderma, nonepidermolytic, 600962 (3)	KRT16	148067	17q12-q21	Dermatological
1176	Palmoplantar verrucous nevus, unilateral, 144200 (3)	KRT16	148067	17q12-q21	Dermatological
3178	Pancreatic agenesis, 260370 (3)	IPF1	600733	13q12.1	Gastrointestinal
1178	Pancreatic cancer, 260350 (3)	ARMET, ARP	601916	3p21.1	Cancer
1178	Pancreatic cancer, 260350 (3)	BRCA2, FANCD1	600185	13q12.3	Cancer
1178	Pancreatic cancer, 260350 (3)	TP53, P53, LFS1	191170	17p13.1	Cancer
1178	Pancreatic cancer (3)	MADH4, DPC4, SMAD4, JIP	600993	18q21.1	Cancer
1178	Pancreatic cancer/melanoma syndrome, 606719 (3)	CDKN2A, MTS1, P16, MLM, CMM2	600160	9p21	Cancer
1178	Pancreatic cancer, somatic (3)	ACVR1B, ACVRLK4, ALK4	601300	12q13	Cancer
1178	Pancreatic cancer, sporadic (3)	STK11, PJS, LKB1	602216	19p13.3	Cancer
1178	Pancreatic carcinoma, somatic, 260350 (3)	KRAS2, RASK2	190070	12p12.1	Cancer
1178	Pancreatic carcinoma, somatic (3)	RBBP8, RIM	604124	18q11.2	Cancer
1179	Pancreatitis, hereditary, 167800 (3)	PRSS1, TRY1	276000	7q35	Gastrointestinal
1179	Pancreatitis, hereditary, 167800 (3)	SPINK1, PSTI, PCTT, TATI	167790	5q32	Gastrointestinal
1179	Pancreatitis, idiopathic (3)	CFTR, ABCC7, CF, MRP7	602421	7q31.2	Gastrointestinal
1183	Papillary serous carcinoma of the peritoneum (3)	BRCA1, PSCP	113705	17q21	Cancer
1184	Papillon-Lefevre syndrome, 245000 (3)	CTSC, CPPI, PALS, PLS, HMS	602365	11q14.1-q14.3	multiple
1186	Paraganglioma, familial malignant, 168000 (3)	SDHB, SDH1, SDHIP	185470	1p36.1-p35	Cancer
1186	Paragangliomas, familial central nervous system, 168000 (3)	SDHD, PGL1	602690	11q23	Cancer
1186	Paragangliomas, familial nonchromaffin, 1, with and without deafness, 168000 (3)	SDHD, PGL1	602690	11q23	Cancer
1186	Paragangliomas, familial nonchromaffin, 3, 605373 (3)	SDHC, PGL3	602413	1q21	Cancer
1186	Paraganglioma, sporadic corotid body, 168000 (3)	SDHD, PGL1	602690	11q23	Cancer
1188	Paramyotonia congenita, 168300 (3)	SCN4A, HYPP, NAC1A	603967	17q23.1-q25.3	Muscular
1189	Parathyroid adenoma, sporadic (3)	MEN1	131100	11q13	Cancer
1189	Parathyroid adenoma with cystic changes, 145001 (3)	HRPT2, C1orf28	607393	1q25-q31	Cancer
1189	Parathyroid carcinoma, 608266 (3)	HRPT2, C1orf28	607393	1q25-q31	Cancer
1190	Parietal foramina 1, 168500 (3)	MSX2, CRS2, HOX8	123101	5q34-q35	Skeletal
1190	Parietal foramina 2, 168500 (3)	ALX4, PFM2, FPP	605420	11p11.2	Skeletal
1190	Parietal foramina with cleidocranial dysplasia, 168550 (3)	MSX2, CRS2, HOX8	123101	5q34-q35	Skeletal
1191	Parkes Weber syndrome, 608355 (3)	RASA1, GAP, CMAVM, PKWS	139150	5q13.3	multiple
1192	Parkinson disease, 168600 (3)	NR4A2, NURR1, NOT, TINUR	601828	2q22-q23	Neurological
1192	Parkinson disease, 168600 (3)	SNCAIP	603779	5q23.1-q23.3	Neurological
1192	Parkinson disease, 168600 (3)	TBP, SCA17	600075	6q27	Neurological
1192	Parkinson disease 4, autosomal dominant Lewy body, 605543 (3)	SNCA, NACP, PARK1, PARK4	163890	4q21	Neurological
1192	Parkinson disease 7, autosomal recessive early-onset, 606324 (3)	DJ1, PARK7	602533	1p36	Neurological
1192	Parkinson disease-8, 607060 (3)	LRRK2, PARK8	609007	12q12	Neurological
1192	Parkinson disease, early onset, 605909 (3)	PINK1, PARK6	608309	1p36	Neurological
1192	Parkinson disease, familial, 168600 (3)	UCHL1, PARK5	191342	4p14	Neurological
1192	Parkinson disease, familial, 168601 (3)	SNCA, NACP, PARK1, PARK4	163890	4q21	Neurological
1192	Parkinson disease, juvenile, type 2, 600116 (3)	PRKN, PARK2, PDJ	602544	6q25.2-q27	Neurological
1192	Parkinson disease, resistance to, 168600 (3)	DBH	609312	9q34	Neurological
1192	Parkinson disease, susceptibility to, 168600 (3)	NDUFV2	600532	18p11.31-p11.2	Neurological
3195	Paroxysmal nocturnal hemoglobinuria (3)	PIGA	311770	Xp22.1	Hematological

1195	Paroxysmal nonkinesigenic dyskinesia, 118800 (3)	MR1, TAHCCP2, KIPP1184, BRP17, PNKD, FPD1, PDC, DYT8	609023	2q35	Neurological
1196	Partington syndrome, 309510 (3)	ARX, ISSX, PRTS, MRXS1, MRX36, MRX54	300382	Xp22.13	Neurological
1198	PCWH, 609136 (3)	SOX10, WS4	602229	22q13	Neurological
1199	Pelger-Huet anomaly, 169400 (3)	LBR, PHA	600024	1q42.1	Hematological
1200	Pelizaeus-Merzbacher disease, 312080 (3)	PLP1, PMD	300401	Xq22	Neurological
1201	Pelizaeus-Merzbacher-like disease, autosomal recessive, 608804 (3)	GJA12, CX47, PMLDAR	608803	1q41-q42	Neurological
1204	Pendred syndrome, 274600 (3)	SLC26A4, PDS, DFNB4	605646	7q31	Ear,Nose,Throat
1205	Perineal hypospadias (3)	AR, DHTR, TFM, SBMA, KD, SMAX1	313700	Xq11-q12	Endocrine
1206	Periodic fever, familial, 142680 (3)	TNFRSF1A, TNFR1, TNFAR, FPF	191190	12p13.2	Immunological
1207	Periodontitis, juvenile, 170650 (3)	CTSC, CPPI, PALS, PLS, HMS	602365	11q14.1-q14.3	Ear,Nose,Throat
1209	Periventricular heterotopia with microcephaly, 608097 (3)	ARFGEF2, BIG2	605371	20q13.13	Neurological
1210	Peroxisomal biogenesis disorder, complementation group 4 (3)	PEX6, PXAAA1, PAF2	601498	6p21.1	multiple
1210	Peroxisomal biogenesis disorder, complementation group 6 (3)	PEX6, PXAAA1, PAF2	601498	6p21.1	multiple
1210	Peroxisome biogenesis factor 12 (3)	PEX12	601758	Chr.17	multiple
3212	Persistent hyperinsulinemic hypoglycemia of infancy, 256450 (3)	KCNJ11, BIR, PHHI	600937	11p15.1	Metabolic
1212	Persistent Mullerian duct syndrome, type I, 261550 (3)	AMH, MIF	600957	19p13.3-p13.2	Developmental
1212	Persistent Mullerian duct syndrome, type II, 261550 (3)	AMHR2, AMHR	600956	12q13	Developmental
1213	Peters anomaly, 603807 (3)	PAX6, AN2, MGDA	607108	11p13	Developmental
1213	Peters anomaly, 604229 (3)	CYP1B1, GLC3A	601771	2p22-p21	Developmental
1214	Peutz-Jeghers syndrome, 175200 (3)	STK11, PJS, LKB1	602216	19p13.3	Cancer
1215	Pfeiffer syndrome, 101600 (3)	FGFR1, FLT2, KAL2	136350	8p11.2-p11.1	Skeletal
1215	Pfeiffer syndrome, 101600 (3)	FGFR2, BEK, CFD1, JWS	176943	10q26	Skeletal
1216	Phenylketonuria (3)	PAH, PKU1	261600	12q24.1	Metabolic
1216	Phenylketonuria due to dihydropteridine reductase deficiency (3)	QDPR, DHPR	261630	4p15.31	Metabolic
1216	Phenylketonuria due to PTS deficiency (3)	PTS	261640	11q22.3-q23.3	Metabolic
1217	Phenylthiocarbamide tasting, 171200 (3)	TAS2R38, T2R61, PTC	607751	7q35-q36	Ear,Nose,Throat
1218	Pheochromocytoma, 171300 (3)	SDHD, PGL1	602690	11q23	Cancer
1218	Pheochromocytoma, 171300 (3)	VHL	608537	3p26-p25	Cancer
1218	Pheochromocytoma, extraadrenal, and cervical paraganglioma, 115310 (3)	SDHB, SDH1, SDHIP	185470	1p36.1-p35	Cancer
1220	Phosphoglycerate dehydrogenase deficiency, 601815 (3)	PHGDH	606879	1q12	Metabolic
1221	Phosphoribosyl pyrophosphate synthetase-related gout (3)	PRPS1	311850	Xq22-q24	Metabolic
1222	Phosphorylase kinase deficiency of liver and muscle, autosomal recessive, 261750 (3)	PHKB	172490	16q12-q13	Metabolic
1223	Phosphoserine phosphatase deficiency (3)	PSP	172480	7p15.2-p15.1	Metabolic
1225	Pick disease, 172700 (3)	PSEN1, AD3	104311	14q24.3	Neurological
1226	Piebaldism (3)	KIT, PBT	164920	4q12	Dermatological
1227	Pigmentation of hair, skin, and eyes, variation in (3)	MATP, AIM1	606202	5p13.3	Dermatological
3229	Pigmented adrenocortical disease, primary isolated, 160980 (3)	PRKAR1A, TSE1, CNC1, CAR	188830	17q23-q24	Cancer
1229	Pigmented paravenous chorioretinal atrophy, 172870 (3)	CRB1, RP12	604210	1q31-q32.1	Ophthamological
1230	Pilomatricoma, 132600 (3)	CTNNB1	116806	3p22-p21.3	Cancer
1232	Pituitary ACTH-secreting adenoma (3)	GNAI2, GNAI2B, GIP	139360	3p21	Cancer
1232	Pituitary ACTH secreting adenoma (3)	GNAS, GNAS1, GPSA, POH, PHP1B, PHP1A, AHO	139320	20q13.2	Cancer
1232	Pituitary adenoma, nonfunctioning (3)	THRA, ERBA1, THRA1	190120	17q11.2	Cancer
3232	Pituitary anomalies with holoprosencephaly-like features (3)	GLI2	165230	2q14	multiple
5232	Pituitary hormone deficiency, combined (3)	POU1F1, PIT1	173110	3p11	Endocrine
5232	Pituitary hormone deficiency, combined (3)	PROP1	601538	5q	Endocrine
5232	Pituitary hormone deficiency, combined, HESX1-related, 182230 (3)	HESX1, RPX	601802	3p21.2-p21.1	Endocrine

5232	Pituitary hormone deficiency, combined, with rigid cervical spine, 262600 (3)	LHX3	600577	9q34.3	Endocrine
7232	Pituitary tumor, invasive (3)	PRKCA, PKCA	176960	17q22-q23.2	Cancer
1233	Placental abruption (3)	NOS3	163729	7q36	Unclassified
5233	Placental steroid sulfatase deficiency (3)	STS, ARSC1, ARSC, SSDD	308100	Xp22.32	Metabolic
1234	Plasmin inhibitor deficiency (3)	PLI, SERPINF2	262850	17pter-p12	Hematological
1235	Plasminogen Tochigi disease (3)	PLG	173350	6q26	Hematological
1237	Platelet-activating factor acetylhydrolase deficiency (3)	PLA2G7, PAFAH	601690	6p21.2-p12	Hematological
1237	Platelet ADP receptor defect (3)	P2RY12, P2Y12	600515	3q24-q25	Hematological
1237	Platelet disorder, familial, with associated myeloid malignancy, 601399 (3)	RUNX1, CBFA2, AML1	151385	21q22.3	Hematological
1237	Platelet glycoprotein IV deficiency, 608404 (3)	CD36	173510	7q11.2	Hematological
1238	Pneumonitis, desquamative interstitial, 263000 (3)	SFTPC, SFTP2	178620	8p21	Respiratory
1239	Pneumothorax, primary spontaneous, 173600 (3)	FLCN, BHD	607273	17p11.2	Respiratory
1241	Polycystic kidney and hepatic disease, 263200 (3)	FCYT, PKHD1, ARPKD	606702	6p21.1-p12	Renal
1241	Polycystic kidney disease, adult type I, 173900 (3)	PKD1	601313	16p13.3-p13.12	Renal
1241	Polycystic kidney disease, adult, type II (3)	PKD2, PKD4	173910	4q21-q23	Renal
1241	Polycystic kidney disease, infantile severe, with tuberous sclerosis (3)	PKDTS	600273	16p13.3	Renal
3241	Polycystic liver disease, 174050 (3)	PRKCSH, G19P1, PCLD	177060	19p13.2-p13.1	Gastrointestinal
3241	Polycystic liver disease, 174050 (3)	SEC63	608648	6q21	Gastrointestinal
1242	Polycythemia, benign familial, 263400 (3)	VHL	608537	3p26-p25	Hematological
1242	Polycythemia vera, 263300 (3)	JAK2	147796	9p24	Hematological
1243	Polydactyly, postaxial, types A1 and B, 174200 (3)	GLI3, PAPA, PAPB, ACLS	165240	7p13	Skeletal
1243	Polydactyly, preaxial, type IV, 174700 (3)	GLI3, PAPA, PAPB, ACLS	165240	7p13	Skeletal
1244	Polymicrogyria, bilateral frontoparietal, 606854 (3)	GPR56, TM7XN1, BFPP	604110	16q13	Neurological
1245	Polyposis, juvenile intestinal, 174900 (3)	BMPR1A, ACVRLK3, ALK3	601299	10q22.3	Cancer
1245	Polyposis, juvenile intestinal, 174900 (3)	MADH4, DPC4, SMAD4, JIP	600993	18q21.1	Cancer
1246	Popliteal pterygium syndrome, 119500 (3)	IRF6, VWS, LPS, PIT, PPS, OFC6	607199	1q32-q41	multiple
1247	Porencephaly, 175780 (3)	COL4A1	120130	13q34	Neurological
1249	Porphyria, acute hepatic (3)	ALAD	125270	9q34	Metabolic
1249	Porphyria, acute intermittent (3)	HMBS, PBGD, UPS	176000	11q23.3	Metabolic
1249	Porphyria, acute intermittent, nonerythroid variant (3)	HMBS, PBGD, UPS	176000	11q23.3	Metabolic
1249	Porphyria, congenital erythropoietic, 263700 (3)	UROS	606938	10q25.2-q26.3	Metabolic
1249	Porphyria cutanea tarda (3)	UROD	176100	1p34	Metabolic
1249	Porphyria, hepatoerythropoietic (3)	UROD	176100	1p34	Metabolic
1249	Porphyria variegata, 176200 (3)	HFE, HLA-H, HFE1	235200	6p21.3	Metabolic
1249	Porphyria variegata, 176200 (3)	PPOX	600923	1q22	Metabolic
1253	PPM-X syndrome, 300055 (3)	MECP2, RTT, PPMX, MRX16, MRX79	300005	Xq28	Neurological
1254	Prader-Willi syndrome, 176270 (3)	NDN	602117	15q11-q13	multiple
1254	Prader-Willi syndrome, 176270 (3)	SNRPN	182279	15q12	multiple
1256	Precocious puberty, male, 176410 (3)	LHCGR	152790	2p21	Developmental
1257	Preeclampsia/eclampsia 4 (3)	STOX1, PEE4	609397	10q22.1	Cardiovascular
1257	Preeclampsia, susceptibility to, 189800 (3)	EPHX1	132810	1q42.1	Cardiovascular
1257	Preeclampsia, susceptibility to (3)	AGT, SERPINA8	106150	1q42-q43	Cardiovascular
1259	Prekallikrein deficiency (3)	KLKB1, KLK3	229000	4q35	Hematological
3260	Premature chromosome condensation with microcephaly and mental retardation, 606858 (3)	MCPH1	607117	8p23	Neurological
1260	Premature ovarian failure, 300511 (3)	DIAPH2, DIA, POF2	300108	Xq22	Renal
1260	Premature ovarian failure 3, 608996 (3)	FOXL2, BPES, BPES1, PFRK, POF3	605597	3q23	Renal
1261	Primary lateral sclerosis, juvenile, 606353 (3)	ALS2, ALSJ, PLSJ, IAHSP	606352	2q33	Neurological
1263	Prion disease with protracted course, 606688 (3)	PRNP, PRIP	176640	20pter-p12	Neurological

1265	Progressive external ophthalmoplegia with mitochondrial DNA deletions, 157640 (3)	C10orf2, TWINKLE, PEO1, PEO	606075	10q24	Ophthamological
1265	Progressive external ophthalmoplegia with mitochondrial DNA deletions, 157640 (3)	POLG, POLG1, POLGA, PEO	174763	15q25	Ophthamological
1265	Progressive external ophthalmoplegia with mitochondrial DNA deletions, 157640 (3)	SLC25A4, ANT1, T1, PEO3	103220	4q35	Ophthamological
1266	Proguanil poor metabolizer (3)	CYP2C, CYP2C19	124020	10q24.1-q24.3	Metabolic
1267	Prolactinoma, hyperparathyroidism, carcinoid syndrome (3)	MEN1	131100	11q13	Endocrine
1268	Prolidase deficiency (3)	PEPD	170100	19cen-q13.11	Connective tissue
1270	Properdin deficiency, X-linked, 312060 (3)	PFC, PFD	300383	Xp11.4-p11.23	Immunological
1271	Propionicacidemia, 606054 (3)	PCCA	232000	13q32	Metabolic
1271	Propionicacidemia, 606054 (3)	PCCB	232050	3q21-q22	Metabolic
1272	Prostate cancer 1, 176807, 601518 (3)	RNASEL, RNS4, PRCA1, HPC1	180435	1q25	Cancer
1272	Prostate cancer, 176807 (3)	BRCA2, FANCD1	600185	13q12.3	Cancer
1272	Prostate cancer, 176807 (3)	PTEN, MMAC1	601728	10q23.31	Cancer
1272	Prostate cancer (3)	AR, DHTR, TFM, SBMA, KD, SMAX1	313700	Xq11-q12	Cancer
1272	Prostate cancer, familial, 176807 (3)	CHEK2, RAD53, CHK2, CDS1, LFS2	604373	22q12.1	Cancer
1272	Prostate cancer, hereditary, 176807 (3)	MSR1	153622	8p22	Cancer
1272	Prostate cancer, progression and metastasis of, 176807 (3)	EPHB2, EPHT3, DRT, ERK	600997	1p36.1-p35	Cancer
1272	Prostate cancer, somatic, 176807 (3)	KLF6, COPEB, BCD1, ZF9	602053	10p15	Cancer
1272	Prostate cancer, somatic, 176807 (3)	MAD1L1, TXBP181	602686	7p22	Cancer
1272	Prostate cancer, susceptibility to, 176807 (3)	AR, DHTR, TFM, SBMA, KD, SMAX1	313700	Xq11-q12	Cancer
1272	Prostate cancer, susceptibility to, 176807 (3)	ATBF1	104155	16q22.3-q23.1	Cancer
1272	Prostate cancer, susceptibility to, 176807 (3)	ELAC2, HPC2	605367	17p11	Cancer
1272	Prostate cancer, susceptibility to, 176807 (3)	MXI1	600020	10q25	Cancer
1273	Protein S deficiency (3)	PROS1	176880	3p11.1-q11.2	Hematological
1274	Proteinuria, low molecular weight, with hypercalciuric nephrocalcinosis (3)	CLCN5, CLCK2, NPHL2, DENTS	300008	Xp11.22	Renal
1276	Protoporphyria, erythropoietic (3)	FECH, FCE	177000	18q21.3	Metabolic
1276	Protoporphyria, erythropoietic, recessive, with liver failure (3)	FECH, FCE	177000	18q21.3	Metabolic
1277	Proud syndrome, 300004 (3)	ARX, ISSX, PRTS, MRXS1, MRX36, MRX54	300382	Xp22.13	multiple
1278	Pseudoachondroplasia, 177170 (3)	COMP, EDM1, MED, PSACH	600310	19p13.1	Skeletal
1279	Pseudohermaphroditism, male, with gynecomastia, 264300 (3)	HSD17B3, EDH17B3	605573	9q22	Developmental
1279	Pseudohermaphroditism, male, with Leydig cell hypoplasia (3)	LHCGR	152790	2p21	Developmental
1281	Pseudohypoaldosteronism, type I, 264350 (3)	SCNN1A	600228	12p13	Endocrine
1281	Pseudohypoaldosteronism, type I, 264350 (3)	SCNN1B	600760	16p13-p12	Endocrine
1281	Pseudohypoaldosteronism, type I, 264350 (3)	SCNN1G, PHA1	600761	16p13-p12	Endocrine
1281	Pseudohypoaldosteronism type I, autosomal dominant, 177735 (3)	NR3C2, MLR, MCR	600983	4q31.1	Endocrine
1281	Pseudohypoaldosteronism type II (3)	WNK4, PRKWNK4, PHA2B	601844	17q21-q22	Endocrine
1281	Pseudohypoaldosteronism, type IIC, 145260 (3)	WNK1, PRKWNK1, KDP, PHA2C	605232	12p13	Endocrine
1282	Pseudohypoparathyroidism, type Ia, 103580 (3)	GNAS, GNAS1, GPSA, POH, PHP1B, PHP1A, AHO	139320	20q13.2	Endocrine
1282	Pseudohypoparathyroidism, type Ib, 603233 (3)	GNAS, GNAS1, GPSA, POH, PHP1B, PHP1A, AHO	139320	20q13.2	Endocrine
1283	Pseudovaginal perineoscrotal hypospadias, 264600 (3)	SRD5A2	607306	2p23	Unclassified
1284	Pseudovitamin D deficiency rickets 1 (3)	CYP27B1, PDDR, VDD1	264700	12q14	Bone
1285	Pseudoxanthoma elasticum, autosomal dominant, 177850 (3)	ABCC6, ARA, ABC34, MLP1, PXE	603234	16p13.1	Connective tissue
1285	Pseudoxanthoma elasticum, autosomal recessive, 264800 (3)	ABCC6, ARA, ABC34, MLP1, PXE	603234	16p13.1	Connective tissue
1288	Psoriasis, susceptibility to, 177900 (3)	PSORS6	605364	19p13	Dermatological

1288	Psoriatic arthritis, susceptibility to, 607507 (3)	CARD15, NOD2, IBD1, CD, ACUG, PSORAS1	605956	16q12	Dermatological
1291	Pulmonary alveolar proteinosis, 265120 (3)	CSF2RB	138981	22q12.2-q13.1	Respiratory
1291	Pulmonary alveolar proteinosis, 265120 (3)	SFTPC, SFTP2	178620	8p21	Respiratory
1291	Pulmonary alveolar proteinosis, congenital, 265120 (3)	SFTPB, SFTB3	178640	2p12-p11.2	Respiratory
1291	Pulmonary fibrosis, idiopathic, familial, 178500 (3)	SFTPC, SFTP2	178620	8p21	Respiratory
1291	Pulmonary fibrosis, idiopathic, susceptibility to, 178500 (3)	SFTPA1, SFTP1	178630	10q22.2-q23.1	Respiratory
5291	Pulmonary hypertension, familial primary, 178600 (3)	BMPR2, PPH1	600799	2q33	Cardiovascular
1293	Pycnodysostosis, 265800 (3)	CTSK	601105	1q21	Skeletal
1294	Pyloric stenosis, infantile hypertrophic, susceptibility to, 179010 (3)	NOS1	163731	12q24.2-q24.31	Developmental
1295	Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416 (3)	PSTPIP1, PSTPIP, CD2BP1, PAPAS	606347	15q24-q25.1	Dermatological
1296	Pyropoikilocytosis (3)	SPTA1	182860	1q21	Hematological
1297	Pyruvate carboxylase deficiency, 266150 (3)	PC	608786	11q13.4-q13.5	Metabolic
1297	Pyruvate dehydrogenase deficiency (3)	PDHA1, PHE1A	312170	Xp22.2-p22.1	Metabolic
1297	Pyruvate dehydrogenase E1-beta deficiency (3)	PDHB	179060	3p13-q23	Metabolic
1298	Rabson-Mendenhall syndrome, 262190 (3)	INSR	147670	19p13.2	multiple
1301	Radioulnar synostosis with amegakaryocytic thrombocytopenia, 605432 (3)	HOXA11, HOX1I	142958	7p15-p14.2	multiple
1303	RAPADILINO syndrome, 266280 (3)	RECQL4, RTS, RECQ4	603780	8q24.3	multiple
1304	Rapid progression to AIDS from HIV1 infection (3)	CX3CR1, GPR13, V28	601470	3pter-p21	Immunological
1305	Rapp-Hodgkin syndrome, 129400 (3)	TP73L, TP63, KET, EEC3, SHFM4, LMS, RHS	603273	3q27	multiple
1306	Red hair/fair skin (3)	MC1R	155555	16q24.3	Dermatological
1307	Refsum disease, 266500 (3)	PEX7, RCDP1	601757	6q22-q24	Neurological
1307	Refsum disease, 266500 (3)	PHYH, PAHX	602026	10pter-p11.2	Neurological
1307	Refsum disease, infantile, 266510 (3)	PEX1, ZWS1	602136	7q21-q22	Neurological
1307	Refsum disease, infantile form, 266510 (3)	PEX26	608666	22q11.21	Neurological
1307	Refsum disease, infantile form, 266510 (3)	PXMP3, PAF1, PMP35, PEX2	170993	8q21.1	Neurological
1308	Renal carcinoma, chromophobe, somatic, 144700 (3)	FLCN, BHD	607273	17p11.2	Cancer
1308	Renal cell carcinoma, 144700 (3)	TRC8, RCA1, HRCA1	603046	8q24.1	Cancer
1308	Renal cell carcinoma, clear cell, somatic, 144700 (3)	OGG1	601982	3p26.2	Cancer
1308	Renal cell carcinoma, papillary, 1, 605074 (3)	PRCC, RCCP1	179755	1q21	Cancer
1308	Renal cell carcinoma, papillary, 1, 605074 (3)	TFE3	314310	Xp11.22	Cancer
1308	Renal cell carcinoma, papillary, familial and sporadic, 605074 (3)	MET	164860	7q31	Cancer
1308	Renal cell carcinoma, somatic (3)	VHL	608537	3p26-p25	Cancer
3308	Renal glucosuria, 233100 (3)	SLC5A2, SGLT2	182381	16p11.2	Renal
5308	Renal hypoplasia, isolated (3)	PAX2	167409	10q24.3-q25.1	Renal
7308	Renal tubular acidosis, distal, 179800, 602722 (3)	SLC4A1, AE1, EPB3	109270	17q21-q22	Renal
7308	Renal tubular acidosis, distal, autosomal recessive, 602722 (3)	ATP6V0A4, ATP6N1B, VPP2, RTA1C, RTADR	605239	7q33-q34	Renal
7308	Renal tubular acidosis-osteopetrosis syndrome (3)	CA2	259730	8q22	Renal
7308	Renal tubular acidosis, proximal, with ocular abnormalities, 604278 (3)	SLC4A4, NBC1, KNBC, SLC4A5	603345	4q21	Renal
7308	Renal tubular acidosis with deafness, 267300 (3)	ATP6B1, VPP3	192132	2cen-q13	Renal
9308	Renal tubular dysgenesis, 267430 (3)	ACE, DCP1, ACE1	106180	17q23	Renal
9308	Renal tubular dysgenesis, 267430 (3)	AGTR1, AGTR1A, AT2R1	106165	3q21-q25	Renal
9308	Renal tubular dysgenesis, 267430 (3)	AGT, SERPINA8	106150	1q42-q43	Renal
9308	Renal tubular dysgenesis, 267430 (3)	REN	179820	1q32	Renal
1309	Renpenning syndrome, 309500 (3)	PQBP1, NPW38, SHS, MRX55, MRXS3, RENS1, MRXS8	300463	Xp11.23	Neurological
1311	Response to morphine-6-glucuronide (3)	OPRM1	600018	6q24-q25	Neurological
1312	Resting heart rate, 607276 (3)	ADRB1, ADRB1R, RHR	109630	10q24-q26	Cardiovascular

1314	Restrictive dermopathy, lethal, 275210 (3)	ZMPSTE24, FACE1, STE24, MADB	606480	1p34	Dermatological
1315	Retinal degeneration, autosomal recessive, clumped pigment type (3)	NRL, D14S46E, RP27	162080	14q11.1-q11.2	Ophthamological
1315	Retinal degeneration, autosomal recessive, prominin-related (3)	PROM1, PROML1, AC133	604365	4p16.2-p12	Ophthamological
1315	Retinal degeneration, late-onset, autosomal dominant, 605670 (3)	C1QTNF5, CTRP5, LORD	608752	11q23.3	Ophthamological
1315	Retinal dystrophy, early-onset severe (3)	LRAT	604863	4q31	Ophthamological
1316	Retinitis pigmentosa-10, 180105 (3)	IMPDH1	146690	7q31.3-q32	Ophthamological
1316	Retinitis pigmentosa-11, 600138 (3)	PRPF31, PRP31	606419	19q13.4	Ophthamological
1316	Retinitis pigmentosa-1, 180100 (3)	RP1, ORP1	603937	8q11-q13	Ophthamological
1316	Retinitis pigmentosa-12, autosomal recessive, 600105 (3)	CRB1, RP12	604210	1q31-q32.1	Ophthamological
1316	Retinitis pigmentosa-13, 600059 (3)	PRPF8, PRPC8, RP13	607300	17p13.3	Ophthamological
1316	Retinitis pigmentosa-14, 600132 (3)	TULP1, RP14	602280	6p21.3	Ophthamological
1316	Retinitis pigmentosa-17, 600852 (3)	CA4, RP17	114760	17q23	Ophthamological
1316	Retinitis pigmentosa-18, 601414 (3)	HPRP3, RP18	607301	1q21.2	Ophthamological
1316	Retinitis pigmentosa-19, 601718 (3)	ABCA4, ABCR, STGD1, FFM, RP19	601691	1p21-p13	Ophthamological
1316	Retinitis pigmentosa-20 (3)	RPE65, RP20	180069	1p31	Ophthamological
1316	Retinitis pigmentosa-2 (3)	RP2	312600	Xp11.3	Ophthamological
1316	Retinitis pigmentosa-26, 608380 (3)	CERKL	608381	2q31.2-q32.3	Ophthamological
1316	Retinitis pigmentosa-27 (3)	NRL, D14S46E, RP27	162080	14q11.1-q11.2	Ophthamological
1316	Retinitis pigmentosa-30, 607921 (3)	FSCN2, RFSN	607643	17q25	Ophthamological
1316	Retinitis pigmentosa-3, 300389 (3)	RPGR, RP3, CRD, RP15, COD1	312610	Xp21.1	Ophthamological
1316	Retinitis pigmentosa-4, autosomal dominant (3)	RHO, RP4, OPN2	180380	3q21-q24	Ophthamological
1316	Retinitis pigmentosa-7, 608133 (3)	RDS, RP7, PRPH2, PRPH, AVMD, AOFMD	179605	6p21.1-cen	Ophthamological
1316	Retinitis pigmentosa-9, 180104 (3)	RP9	607331	7p14.2	Ophthamological
1316	Retinitis pigmentosa, AR, 268000 (3)	RLBP1	180090	15q26	Ophthamological
1316	Retinitis pigmentosa, AR, without hearing loss, 268000 (3)	USH2A	608400	1q41	Ophthamological
1316	Retinitis pigmentosa, autosomal dominant (3)	RGR	600342	10q23	Ophthamological
1316	Retinitis pigmentosa, autosomal recessive, 268000 (3)	CNGB1, CNCG3L, CNCG2	600724	16q13	Ophthamological
1316	Retinitis pigmentosa, autosomal recessive (3)	CNGA1, CNCG1	123825	4p12-cen	Ophthamological
1316	Retinitis pigmentosa, autosomal recessive (3)	PDE6A, PDEA	180071	5q31.2-q34	Ophthamological
1316	Retinitis pigmentosa, autosomal recessive (3)	PDE6B, PDEB, CSNB3	180072	4p16.3	Ophthamological
1316	Retinitis pigmentosa, autosomal recessive (3)	RGR	600342	10q23	Ophthamological
1316	Retinitis pigmentosa, autosomal recessive (3)	RHO, RP4, OPN2	180380	3q21-q24	Ophthamological
1316	Retinitis pigmentosa, digenic (3)	ROM1, ROSP1	180721	11q13	Ophthamological
1316	Retinitis pigmentosa, digenic, 608133 (3)	RDS, RP7, PRPH2, PRPH, AVMD, AOFMD	179605	6p21.1-cen	Ophthamological
1316	Retinitis pigmentosa, juvenile (3)	AIPL1, LCA4	604392	17p13.1	Ophthamological
1316	Retinitis pigmentosa, late onset, 268000 (3)	NR2E3, PNR, ESCS	604485	15q23	Ophthamological
1316	Retinitis pigmentosa, late-onset dominant, 268000 (3)	CRX, CORD2, CRD	602225	19q13.3	Ophthamological
1316	Retinitis pigmentosa, MERTK-related, 268000 (3)	MERTK	604705	2q14.1	Ophthamological
1316	Retinitis pigmentosa, X-linked with deafness and sinorespiratory infections, 300455 (3)	RPGR, RP3, CRD, RP15, COD1	312610	Xp21.1	Ophthamological
1316	Retinitis pigmentosa, X-linked, with recurrent respiratory infections, 300455 (3)	RPGR, RP3, CRD, RP15, COD1	312610	Xp21.1	Ophthamological
1316	Retinitis punctata albescens, 136880 (3)	RDS, RP7, PRPH2, PRPH, AVMD, AOFMD	179605	6p21.1-cen	Ophthamological
1316	Retinitis punctata albescens, 136880 (3)	RLBP1	180090	15q26	Ophthamological
1317	Retinoblastoma (3)	RB1	180200	13q14.1-q14.2	Cancer
1318	Retinol binding protein, deficiency of (3)	RBP4	180250	10q24	Ophthamological
1320	Retinoschisis (3)	RS1, XLRS1	312700	Xp22.2-p22.1	Ophthamological
1321	Rett syndrome, 312750 (3)	MECP2, RTT, PPMX, MRX16, MRX79	300005	Xq28	Neurological

1321	Rett syndrome, atypical, 312750 (3)	CDKL5, STK9	300203	Xp22	Neurological
1321	Rett syndrome, preserved speech variant, 312750 (3)	MECP2, RTT, PPMX, MRX16, MRX79	300005	Xq28	Neurological
1322	Rhabdoid predisposition syndrome, familial (3)	SMARCB1, SNF5, INI1, RDT	601607	22q11	Cancer
1322	Rhabdoid tumors (3)	SMARCB1, SNF5, INI1, RDT	601607	22q11	Cancer
1323	Rhabdomyosarcoma, 268210 (3)	SLC22A1L, BWSCR1A, IMPT1	602631	11p15.5	Cancer
1323	Rhabdomyosarcoma, alveolar, 268220 (3)	FOXO1A, FKHR	136533	13q14.1	Cancer
1323	Rhabdomyosarcoma, alveolar, 268220 (3)	PAX3, WS1, HUP2, CDHS	606597	2q35	Cancer
1323	Rhabdomyosarcoma, alveolar, 268220 (3)	PAX7	167410	1p36.2-p36.12	Cancer
1324	Rheumatoid arthritis, progression of, 180300 (3)	IL10, CSIF	124092	1q31-q32	Connective tissue
1324	Rheumatoid arthritis, susceptibility to, 180300 (3)	MHC2TA, C2TA	600005	16p13	Connective tissue
1324	Rheumatoid arthritis, susceptibility to, 180300 (3)	NFKBIL1	601022	6p21.3	Connective tissue
1324	Rheumatoid arthritis, susceptibility to, 180300 (3)	PADI4, PADI5, PAD	605347	1p36	Connective tissue
1324	Rheumatoid arthritis, susceptibility to, 180300 (3)	PTPN8, PEP, PTPN22, LYP	600716	1p13	Connective tissue
1324	Rheumatoid arthritis, susceptibility to, 180300 (3)	RUNX1, CBFA2, AML1	151385	21q22.3	Connective tissue
1324	Rheumatoid arthritis, susceptibility to, 180300 (3)	SLC22A4, OCTN1	604190	5q31	Connective tissue
1324	Rheumatoid arthritis, systemic juvenile, susceptibility to, 604302 (3)	MIF	153620	22q11.2	Connective tissue
1325	Rhizomelic chondrodysplasia punctata, type 1, 215100 (3)	PEX7, RCDP1	601757	6q22-q24	multiple
1325	Rhizomelic chondrodysplasia punctata, type 3, 600121 (3)	AGPS, ADHAPS	603051	2q31	multiple
1326	Rh-mod syndrome (3)	RHAG, RH50A	180297	6p21.1-p11	Hematological
1327	Rh-negative blood type (3)	RHD	111680	1p36.2-p34	Hematological
1327	Rh-null disease, amorph type (3)	RHCE	111700	1p36.2-p34	Hematological
1329	Ribose 5-phosphate isomerase deficiency, 608611 (3)	RPIA, RPI	180430	2p11.2	Metabolic
1330	Rickets due to defect in vitamin D 25-hydroxylation, 600081 (3)	CYP2R1	608713	11p15.2	Bone
1330	Rickets, vitamin D-resistant, type IIA, 277440 (3)	VDR	601769	12q12-q14	Bone
1330	Rickets, vitamin D-resistant, type IIB, 277420 (3)	VDR	601769	12q12-q14	Bone
1331	Rieger anomaly (3)	FOXC1, FKHL7, FREAC3	601090	6p25	multiple
1331	Rieger syndrome, 180500 (3)	PITX2, IDG2, RIEG1, RGS, IGDS2	601542	4q25-q26	multiple
1332	Ring dermoid of cornea, 180550 (3)	PITX2, IDG2, RIEG1, RGS, IGDS2	601542	4q25-q26	Ophthamological
1333	Rippling muscle disease, 606072 (3)	CAV3, LGMD1C	601253	3p25	Muscular
1334	Roberts syndrome, 268300 (3)	ESCO2	609353	8p21.1	Developmental
1335	Robinow syndrome, autosomal recessive, 268310 (3)	ROR2, BDB1, BDB, NTRKR2	602337	9q22	multiple
1337	Rokitansky-Kuster-Hauser syndrome, 277000 (3)	WNT4	603490	1p35	Developmental
1338	Rothmund-Thomson syndrome, 268400 (3)	RECQL4, RTS, RECQ4	603780	8q24.3	multiple
1339	Roussy-Levy syndrome, 180800 (3)	MPZ, CMT1B, CMTDI3, CHM, DSS	159440	1q22	multiple
1339	Roussy-Levy syndrome, 180800 (3)	PMP22, CMT1A, CMT1E, DSS	601097	17p11.2	multiple
1341	Rubenstein-Taybi syndrome, 180849 (3)	CREBBP, CBP, RSTS	600140	16p13.3	multiple
1341	Rubinstein-Taybi syndrome, 180849 (3)	EP300	602700	22q13	multiple
1344	Saethre-Chotzen syndrome, 101400 (3)	FGFR2, BEK, CFD1, JWS	176943	10q26	Developmental
1344	Saethre-Chotzen syndrome, 101400 (3)	TWIST, ACS3, SCS	601622	7p21	Developmental
1344	Saethre-Chotzen syndrome with eyelid anomalies, 101400 (3)	TWIST, ACS3, SCS	601622	7p21	Developmental
1345	Salivary adenoma (3)	HMGA2, HMGIC, BABL, LIPO	600698	12q14.3	Cancer
1346	Salla disease, 604369 (3)	SLC17A5, SIASD, SLD	604322	6q14-q15	Metabolic
1347	Sandhoff disease, infantile, juvenile, and adult forms, 268800 (3)	HEXB	606873	5q13	Metabolic
1348	Sanfilippo syndrome, type A, 252900 (3)	SGSH, MPS3A, SFMD	605270	17q25.3	Metabolic
1348	Sanfilippo syndrome, type B (3)	NAGLU	252920	17q21	Metabolic
1349	Sarcoidosis, early-onset, 181000 (3)	CARD15, NOD2, IBD1, CD, ACUG, PSORAS1	605956	16q12	Immunological
1349	Sarcoidosis, susceptibility to, 181000 (3)	BTNL2	606000	6p21.3	Immunological
1349	Sarcoidosis, susceptibility to, 181000 (3)	HLA-DR1B	142857	6p21.3	Immunological
1350	Sarcoma, synovial (3)	SSX1, SSRC	312820	Xp11.2	Cancer

1350	Sarcoma, synovial (3)	SSX2	300192	Xp11.2	Cancer
1352	SARS, progression of (3)	ACE, DCP1, ACE1	106180	17q23	Immunological
1354	Schimke immunoosseous dysplasia, 242900 (3)	SMARCAL1, HARP, SIOD	606622	2q34-q36	Connective tissue
1355	Schindler disease, type I, 609241 (3)	NAGA	104170	22q11	Metabolic
1355	Schindler disease, type III, 609241 (3)	NAGA	104170	22q11	Metabolic
1357	Schizencephaly, 269160 (3)	EMX2	600035	10q26.1	Neurological
1359	Schizoaffective disorder, susceptibility to, 181500 (3)	DISC1	605210	1q42.1	Psychiatric
1359	Schizophrenia 5, 603175 (3)	TRAR4	608923	6q23.2	Psychiatric
1359	Schizophrenia, chronic (3)	APP, AAA, CVAP, AD1	104760	21q21	Psychiatric
1359	Schizophrenia, susceptibility to, 181500 (3)	COMT	116790	22q11.2	Psychiatric
1359	Schizophrenia, susceptibility to, 181500 (3)	DISC1	605210	1q42.1	Psychiatric
1359	Schizophrenia, susceptibility to, 181500 (3)	HTR2A	182135	13q14-q21	Psychiatric
1359	Schizophrenia, susceptibility to, 181500 (3)	RTN4R, NOGOR	605566	22q11	Psychiatric
1359	Schizophrenia, susceptibility to, 181500 (3)	SYN2	600755	3p25	Psychiatric
1359	Schizophrenia, susceptibility to, 181510 (3)	EPN4, EPNR, KIAA0171, SCZD1	607265	5q33.3	Psychiatric
1359	Schizophrenia, susceptibility to, 4 600850 (3)	PRODH, PRODH2, SCZD4	606810	22q11.2	Psychiatric
1360	Schwannomatosis, 162091 (3)	NF2	607379	22q12.2	Cancer
1361	Schwartz-Jampel syndrome, type 1, 255800 (3)	HSPG2, PLC, SJS, SJA, SJS1	142461	1p36.1	multiple
1362	SCID, autosomal recessive, T-negative/B-positive type (3)	JAK3, JAKL	600173	19p13.1	Immunological
1363	Sclerosteosis, 269500 (3)	SOST	605740	17q12-q21	Skeletal
1365	Scurvy (3)	GULOP, GULO	240400	8p21.1	Nutritional
1366	Sea-blue histiocyte disease, 269600 (3)	APOE, AD2	107741	19q13.2	Hematological
1367	Seasonal affective disorder, susceptibility to, 608516 (3)	HTR2A	182135	13q14-q21	Psychiatric
1368	Sebastian syndrome, 605249 (3)	MYH9, MHA, FTNS, DFNA17	160775	22q11.2	Hematological
1369	Seckel syndrome 1, 210600 (3)	ATR, FRP1, SCKL	601215	3q22-q24	Developmental
1370	Segawa syndrome, recessive (3)	TH, TYH	191290	11p15.5	Neurological
1371	Seizures, afebrile, 604233 (3)	SCN2A1, SCN2A	182390	2q23-q24.3	Neurological
1371	Seizures, benign familial neonatal-infantile, 607745 (3)	SCN2A1, SCN2A	182390	2q23-q24.3	Neurological
1372	Selective T-cell defect (3)	ZAP70, SRK, STD	176947	2q12	Immunological
1373	Self-healing collodion baby, 242300 (3)	TGM1, ICR2, LI1	190195	14q11.2	Dermatological
1374	SEMD, Pakistani type (3)	PAPSS2, ATPSK2	603005	10q22-q24	Connective tissue
1375	Senior-Loken syndrome-1, 266900 (3)	NPHP1, NPH1, SLSN1	607100	2q13	Renal
1375	Senior-Loken syndrome 4, 606996 (3)	NPHP4, SLSN4	607215	1p36	Renal
1375	Senior-Loken syndrome 5, 609254 (3)	IQCB1, NPHP5, KIAA0036	609237	3q21.1	Renal
1376	Sensory ataxic neuropathy, dysarthria, and ophthalmoparesis, 157640 (3)	POLG, POLG1, POLGA, PEO	174763	15q25	Ophthamological
1377	Sepiapterin reductase deficiency (3)	SPR	182125	2p14-p12	Metabolic
1378	Sepsis, susceptibility to (3)	CASP12, CASP12P1	608633	11q22.3	Immunological
1378	Septic shock, susceptibility to (3)	TNF, TNFA	191160	6p21.3	Immunological
1380	Septooptic dysplasia, 182230 (3)	HESX1, RPX	601802	3p21.2-p21.1	multiple
1381	Sertoli cell-only syndrome, susceptibility to, 305700 (3)	USP26	300309	Xq26.2	Renal
1383	Severe combined immunodeficiency, Athabascan type, 602450 (3)	DCLRE1C, ARTEMIS, SCIDA	605988	10p	Immunological
1383	Severe combined immunodeficiency, B cell-negative, 601457 (3)	RAG1	179615	11p13	Immunological
1383	Severe combined immunodeficiency, B cell-negative, 601457 (3)	RAG2	179616	11p13	Immunological
1383	Severe combined immunodeficiency due to ADA deficiency, 102700 (3)	ADA	608958	20q13.11	Immunological
1383	Severe combined immunodeficiency due to PTPRC deficiency (3)	PTPRC, CD45, LCA	151460	1q31-q32	Immunological
1383	Severe combined immunodeficiency, T-cell negative, B-cell/natural killer cell-positive type, 600802 (3)	IL7R	146661	5p13	Immunological
1383	Severe combined immunodeficiency, T-negative/B-positive type, 600802 (3)	CD3D, T3D	186790	11q23	Immunological
1383	Severe combined immunodeficiency, X-linked, 300400 (3)	IL2RG, SCIDX1, SCIDX, IMD4	308380	Xq13	Immunological
1384	Sex reversal, XY, with adrenal failure (3)	FTZF1, FTZ1, SF1	184757	9q33	Unclassified

1385	Sezary syndrome (3)	BCL10	603517	1p22	Cancer
1386	Shah-Waardenburg syndrome, 277580 (3)	EDN3	131242	20q13.2-q13.3	multiple
1387	Short stature, autosomal dominant, with normal serum growth hormone binding protein (3)	GHR	600946	5p13-p12	Skeletal
1387	Short stature, idiopathic (3)	GHR	600946	5p13-p12	Skeletal
1387	Short stature, idiopathic familial, 604271 (3)	SHOX, GCFX, SS, PHOG	312865	Xpter-p22.32	Skeletal
1387	Short stature, idiopathic familial, 604271 (3)	SHOXY	400020	Ypter-p11.2	Skeletal
1387	Short stature, pituitary and cerebellar defects, and small sella turcica, 606606 (3)	LHX4	602146	1q25	Skeletal
1388	Shprintzen-Goldberg syndrome, 182212 (3)	FBN1, MFS1, WMS	134797	15q21.1	multiple
1389	Shwachman-Diamond syndrome, 260400 (3)	SBDS, SDS	607444	7q11	multiple
1391	Sialic acid storage disorder, infantile, 269920 (3)	SLC17A5, SIASD, SLD	604322	6q14-q15	Metabolic
1391	Sialidosis, type I, 256550 (3)	NEU1, NEU, SIAL1	608272	6p21.3	Metabolic
1391	Sialidosis, type II, 256550 (3)	NEU1, NEU, SIAL1	608272	6p21.3	Metabolic
1392	Sialuria, 269921 (3)	GNE, GLCNE, IBM2, DMRV, NM	603824	9p12-p11	Metabolic
1393	Sickle cell anemia (3)	HBB	141900	11p15.5	Hematological
1394	Sick sinus syndrome, 608567 (3)	SCN5A, LQT3, IVF, HB1, SSS1	600163	3p21	Cardiovascular
1396	Silver spastic paraplegia syndrome, 270685 (3)	BSCL2, SPG17	606158	11q13	Neurological
1397	Simpson-Golabi-Behmel syndrome, type 1, 312870 (3)	GPC3, SDYS, SGBS1	300037	Xq26	multiple
1398	Sitosterolemia, 210250 (3)	ABCG5	605459	2p21	Metabolic
1398	Sitosterolemia, 210250 (3)	ABCG8	605460	2p21	Metabolic
1399	Situs ambiguus (3)	NODAL	601265	Chr.10	Developmental
1399	Situs inversus viscerum, 270100 (3)	DNAH11, DNAHC11	603339	Chr.7	Developmental
1400	Sjogren-Larsson syndrome, 270200 (3)	ALDH3A2, ALDH10, SLS, FALDH	609523	17p11.2	Metabolic
1401	Skin fragility-woolly hair syndrome, 607655 (3)	DSP, KPPS2, PPKS2	125647	6p24	Dermatological
1403	Slow acetylation (3)	NAT2, AAC2	243400	8p23.1-p21.3	Metabolic
1404	Slowed nerve conduction velocity, AD, 608236 (3)	ARHGEF10, KIAA0294	608136	8p23	Neurological
1406	Small patella syndrome, 147891 (3)	TBX4	601719	17q21-q22	Skeletal
1408	SMED Strudwick type, 184250 (3)	COL2A1	120140	12q13.11-q13.2	Skeletal
1409	Smith-Fineman-Myers syndrome, 309580 (3)	ATRX, XH2, XNP, MRXS3, SHS	300032	Xq13	multiple
1410	Smith-Lemli-Opitz syndrome, 270400 (3)	DHCR7, SLOS	602858	11q12-q13	multiple
1411	Smith-Magenis syndrome, 182290 (3)	RAI1, SMCR, SMS	607642	17p11.2	multiple
1412	Smith-McCort dysplasia, 607326 (3)	DYM, FLJ90130, DMC, SMC	607461	18q12-q21.1	Skeletal
1414	Solitary median maxillary central incisor, 147250 (3)	SHH, HPE3, HLP3, SMMCI	600725	7q36	Skeletal
1415	Somatotrophinoma (3)	GNAS, GNAS1, GPSA, POH, PHP1B, PHP1A, AHO	139320	20q13.2	Endocrine
1416	Sorsby fundus dystrophy, 136900 (3)	TIMP3, SFD	188826	22q12.1-q13.2	Ophthamological
1417	Sotos syndrome, 117550 (3)	NSD1, ARA267, STO	606681	5q35	Developmental
1418	Spastic ataxia, Charlevoix-Saguenay type, 270550 (3)	SACS, ARSACS	604490	13q12	Neurological
1418	Spastic paralysis, infantile onset ascending, 607225 (3)	ALS2, ALSJ, PLSJ, IAHSP	606352	2q33	Neurological
1418	Spastic paraplegia 10, 604187 (3)	KIF5A, NKHC, SPG10	602821	12q13	Neurological
1418	Spastic paraplegia-13, 605280 (3)	HSPD1, SPG13, HSP60	118190	2q33.1	Neurological
1418	Spastic paraplegia-2, 312920 (3)	PLP1, PMD	300401	Xq22	Neurological
1418	Spastic paraplegia-3A, 182600 (3)	SPG3A	606439	14q11-q21	Neurological
1418	Spastic paraplegia-4, 182601 (3)	SPG4, SPAST	604277	2p22-p21	Neurological
1418	Spastic paraplegia-6, 600363 (3)	NIPA1, SPG6	608145	15q11.1	Neurological
1418	Spastic paraplegia-7, 607259 (3)	PGN, SPG7, CMAR, CAR	602783	16q24.3	Neurological
3419	Specific granule deficiency, 245480 (3)	CEBPE, CRP1	600749	14q11.2	Immunological
1419	Speech-language disorder-1, 602081 (3)	FOXP2, SPCH1, TNRC10, CAGH44	605317	7q31	Neurological
1422	Spermatogenic failure, susceptibility to (3)	DAZL, DAZH, SPGYLA	601486	3p24	Renal

1423	Spherocytosis-1 (3)	SPTB	182870	14q22-q23.2	Hematological
1423	Spherocytosis-2 (3)	ANK1, SPH2	182900	8p11.2	Hematological
1423	Spherocytosis, hereditary (3)	SLC4A1, AE1, EPB3	109270	17q21-q22	Hematological
1423	Spherocytosis, hereditary, Japanese type (3)	EPB42	177070	15q15	Hematological
1423	Spherocytosis, recessive (3)	SPTA1	182860	1q21	Hematological
1425	Spina bifida, 601634 (3)	MTHFD, MTHFC	172460	14q24	Developmental
1425	Spina bifida, risk of, 601634, 182940 (3)	MTR	156570	1q43	Developmental
1425	Spina bifida, risk of, 601634, 182940 (3)	MTRR	602568	5p15.3-p15.2	Developmental
1426	Spinal and bulbar muscular atrophy of Kennedy, 313200 (3)	AR, DHTR, TFM, SBMA, KD, SMAX1	313700	Xq11-q12	Muscular
1426	Spinal muscrular atrophy, late-onset, Finkel type, 182980 (3)	VAPB, VAPC, ALS8	605704	20q13.3	Muscular
1426	Spinal muscular atrophy-1, 253300 (3)	SMN1, SMA1, SMA2, SMA3, SMA4	600354	5q12.2-q13.3	Muscular
1426	Spinal muscular atrophy-2, 253550 (3)	SMN1, SMA1, SMA2, SMA3, SMA4	600354	5q12.2-q13.3	Muscular
1426	Spinal muscular atrophy-3, 253400 (3)	SMN1, SMA1, SMA2, SMA3, SMA4	600354	5q12.2-q13.3	Muscular
1426	Spinal muscular atrophy-4, 271150 (3)	SMN1, SMA1, SMA2, SMA3, SMA4	600354	5q12.2-q13.3	Muscular
1426	Spinal muscular atrophy, distal, type V, 600794 (3)	BSCL2, SPG17	606158	11q13	Muscular
1426	Spinal muscular atrophy, distal, type V, 600794 (3)	GARS, SMAD1, CMT2D	600287	7p15	Muscular
1426	Spinal muscular atrophy, juvenile (3)	HEXB	606873	5q13	Muscular
1426	Spinal muscular atrophy with respiratory distress, 604320 (3)	IGHMBP2, SMUBP2, CATF1, SMARD1	600502	11q13.2-q13.4	Muscular
1428	Spinocerebellar ataxia-10 (3)	ATXN10, SCA10	603516	22q13	Neurological
1428	Spinocerebellar ataxia-1, 164400 (3)	ATXN1, ATX1, SCA1	601556	6p23	Neurological
1428	Spinocerebellar ataxia 12, 604326 (3)	PPP2R2B	604325	5q31-q33	Neurological
1428	Spinocerebellar ataxia 14, 605361 (3)	PRKCG, PKCC, PKCG, SCA14	176980	19q13.4	Neurological
1428	Spinocerebellar ataxia 17, 607136 (3)	TBP, SCA17	600075	6q27	Neurological
1428	Spinocerebellar ataxia-2, 183090 (3)	ATXN2, ATX2, SCA2	601517	12q24	Neurological
1428	Spinocerebellar ataxia 25 (3)	SCA25	608703	2p21-p13	Neurological
1428	Spinocerebellar ataxia-27, 609307 (3)	FGF14, FHF4, SCA27	601515	13q34	Neurological
1428	Spinocerebellar ataxia 4, pure Japanese type, 117210 (3)	PLEKHG4	609526	16q22.1	Neurological
1428	Spinocerebellar ataxia-6, 183086 (3)	CACNA1A, CACNL1A4, SCA6	601011	19p13	Neurological
1428	Spinocerebellar ataxia-7, 164500 (3)	ATXN7, SCA7, OPCA3	607640	3p21.1-p12	Neurological
1428	Spinocerebellar ataxia 8, 608768 (3)	SCA8	603680	13q21	Neurological
1428	Spinocerebellar ataxia, autosomal recessive with axonal neuropathy, 607250 (3)	TDP1	607198	14q31-q32	Neurological
1430	Split hand/foot malformation, type 3, 600095 (3)	SHFM3, DAC	608071	10q24	Skeletal
1430	Split-hand/foot malformation, type 4, 605289 (3)	TP73L, TP63, KET, EEC3, SHFM4, LMS, RHS	603273	3q27	Skeletal
1432	Spondylocarpotarsal synostosis syndrome, 272460 (3)	FLNB, SCT, AOI	603381	3p14.3	Skeletal
1433	Spondylocostal dysostosis, autosomal recessive, 1, 277300 (3)	DLL3, SCDO1	602768	19q13	Skeletal
1433	Spondylocostal dysostosis, autosomal recessive 2, 608681 (3)	MESP2	605195	15q26.1	Skeletal
1435	Spondyloepimetaphyseal dysplasia, 608728 (3)	MATN3, EDM5, HOA	602109	2p24-p23	Skeletal
1435	Spondyloepiphyseal dysplasia, Kimberley type, 608361 (3)	AGC1, CSPG1, MSK16, SEDK	155760	15q26.1	Skeletal
1435	Spondyloepiphyseal dysplasia, Omani type, 608637 (3)	CHST3, C6ST, C6ST1	603799	10q22.1	Skeletal
1435	Spondyloepiphyseal dysplasia tarda, 313400 (3)	SEDL, SEDT	300202	Xp22.2-p22.1	Skeletal
1435	Spondyloepiphyseal dysplasia tarda with progressive arthropathy, 208230 (3)	WISP3, PPAC, PPD	603400	6q22-q23	Skeletal
1436	Spondylometaphyseal dysplasia, Japanese type (3)	COL10A1	120110	6q21-q22.3	Skeletal
1437	Squamous cell carcinoma, burn scar-related, somatic (3)	TNFRSF6, APT1, FAS, CD95, ALPS1A	134637	10q24.1	Cancer
1437	Squamous cell carcinoma, head and neck, 601400 (3)	ING1	601566	13q34	Cancer
1437	Squamous cell carcinoma, head and neck, 601400 (3)	TNFRSF10B, DR5, TRAILR2	603612	8p22-p21	Cancer
1438	Stapes ankylosis syndrome without symphalangism, 184460 (3)	NOG, SYM1, SYNS1	602991	17q22	multiple
1439	Stargardt disease-1, 248200 (3)	ABCA4, ABCR, STGD1, FFM, RP19	601691	1p21-p13	Ophthamological
1439	Stargardt disease 3, 600110 (3)	ELOVL4, ADMD, STGD2, STGD3	605512	6q14	Ophthamological

1440	Startle disease, autosomal recessive (3)	GLRA1, STHE	138491	5q32	Neurological
1440	Startle disease/hyperekplexia, autosomal dominant, 149400 (3)	GLRA1, STHE	138491	5q32	Neurological
1441	STAT1 deficiency, complete (3)	STAT1	600555	2q32.2-q32.3	Unclassified
1442	Statins, attenuated cholesterol lowering by (3)	HMGCR	142910	5q13.3-q14	Metabolic
1444	Steatocystoma multiplex, 184500 (3)	KRT17, PC2, PCHC1	148069	17q12-q21	Dermatological
1445	Stem-cell leukemia/lymphoma syndrome (3)	ZNF198, SCLL, RAMP, FIM	602221	13q11-q12	Cancer
1446	Stevens-Johnson syndrome, carbamazepine-induced, susceptibility to, 608579 (3)	HLA-B	142830	6p21.3	Dermatological
1447	Stickler syndrome, type I, 108300 (3)	COL2A1	120140	12q13.11-q13.2	multiple
1447	Stickler syndrome, type II, 604841 (3)	COL11A1, STL2	120280	1p21	multiple
1447	Stickler syndrome, type III, 184840 (3)	COL11A2, STL3, DFNA13	120290	6p21.3	multiple
1449	Stomach cancer, 137215 (3)	KRAS2, RASK2	190070	12p12.1	Cancer
1454	Stroke, susceptibility to, 1, 606799 (3)	PDE4D, DPDE3, STRK1	600129	5q12	Cardiovascular
1454	Stroke, susceptibility to, 601367 (3)	ALOX5AP, FLAP	603700	13q12	Cardiovascular
1455	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome, 601559 (3)	LIFR, STWS, SWS, SJS2	151443	5p13.1	multiple
1456	Subcortical laminal heteropia, X-linked, 300067 (3)	DCX, DBCN, LISX	300121	Xq22.3-q23	Neurological
1456	Subcortical laminar heterotopia (3)	PAFAH1B1, LIS1	601545	17p13.3	Neurological
1457	Succinic semialdehyde dehydrogenase deficiency (3)	SSADH	271980	6p22	Metabolic
1458	Sucrose intolerance (3)	SI	222900	3q25-q26	Metabolic
1459	Sudden infant death with dysgenesis of the testes syndrome, 608800 (3)	TSPYL1, TSPYL, SIDDT	604714	6q22-q23	Unclassified
1460	Sulfite oxidase deficiency, 272300 (3)	SUOX	606887	Chr.12	Metabolic
1461	Superoxide dismutase, elevated extracellular (3)	SOD3	185490	4p15.3-p15.1	Unclassified
1462	Supranuclear palsy, progressive, 601104 (3)	MAPT, MTBT1, DDPAC, MSTD	157140	17q21.1	Neurological
1462	Supranuclear palsy, progressive atypical, 260540 (3)	MAPT, MTBT1, DDPAC, MSTD	157140	17q21.1	Neurological
1463	Supravalvar aortic stenosis, 185500 (3)	ELN	130160	7q11.2	Cardiovascular
1464	Surfactant deficiency, neonatal, 267450 (3)	ABCA3, ABC3	601615	16p13.3	Respiratory
1464	Surfactant protein C deficiency (3)	SFTPC, SFTP2	178620	8p21	Respiratory
1465	Sutherland-Haan syndrome-like, 300465 (3)	ATRX, XH2, XNP, MRXS3, SHS	300032	Xq13	multiple
1466	Sweat chloride elevation without CF (3)	CFTR, ABCC7, CF, MRP7	602421	7q31.2	Unclassified
1467	Symphalangism, proximal, 185800 (3)	NOG, SYM1, SYNS1	602991	17q22	Skeletal
1468	Syndactyly, type III, 186100 (3)	GJA1, CX43, ODDD, SDTY3, ODOD	121014	6q21-q23.2	Skeletal
1469	Synostoses syndrome, multiple, 1, 186500 (3)	NOG, SYM1, SYNS1	602991	17q22	multiple
1470	Synpolydactyly, 3/3'4, associated with metacarpal and metatarsal synostoses, 608180 (3)	FBLN1	135820	22q13.3	Skeletal
1470	Synpolydactyly, type II, 186000 (3)	HOXD13, HOX4I, SPD	142989	2q31-q32	Skeletal
1470	Synpolydactyly with foot anomalies, 186000 (3)	HOXD13, HOX4I, SPD	142989	2q31-q32	Skeletal
1471	Systemic lupus erythematosus, susceptibility, 152700 (3)	TNFSF6, APT1LG1, FASL	134638	1q23	Immunological
1471	Systemic lupus erythematosus, susceptibility to, 152700 (3)	DNASE1, DNL1	125505	16p13.3	Immunological
1471	Systemic lupus erythematosus, susceptibility to, 152700 (3)	PTPN8, PEP, PTPN22, LYP	600716	1p13	Immunological
1471	Systemic lupus erythematosus, susceptibility to, 2, 605218, 152700 (3)	PDCD1, SLEB2	600244	2q37.3	Immunological
1472	Tall stature, susceptibility to (3)	MCM6	601806	2q21	Skeletal
1473	Tangier disease, 205400 (3)	ABCA1, ABC1, HDLDT1, TGD	600046	9q22-q31	Metabolic
1475	Tarsal-carpal coalition syndrome, 186570 (3)	NOG, SYM1, SYNS1	602991	17q22	Skeletal
1476	Tauopathy and respiratory failure (3)	MAPT, MTBT1, DDPAC, MSTD	157140	17q21.1	Neurological
1477	Tay-Sachs disease, 272800 (3)	HEXA, TSD	606869	15q23-q24	Metabolic
1478	T-cell acute lymphoblastic leukemia (3)	BAX	600040	19q13.3-q13.4	Cancer
3478	T-cell immunodeficiency, congenital alopecia, and nail dystrophy (3)	WHN	600838	17q11-q12	Immunological
1478	T-cell prolymphocytic leukemia, sporadic (3)	ATM, ATA, AT1	607585	11q22.3	Cancer
1480	Temperature-sensitive apoptosis, cellular (3)	DAD1	600243	14q11-q12	Unclassified
1482	Tetra-amelia, autosomal recessive, 273395 (3)	WNT3, INT4	165330	17q21	multiple

1483	Tetralogy of Fallot, 187500 (3)	JAG1, AGS, AHD	601920	20p12	Cardiovascular
1483	Tetralogy of Fallot, 187500 (3)	ZFPM2, FOG2	603693	8q23	Cardiovascular
1483	Tetrology of Fallot, 187500 (3)	NKX2E, CSX	600584	5q34	Cardiovascular
1486	Thalassemia, alpha- (3)	HBA2	141850	16pter-p13.3	Hematological
1486	Thalassemia-beta, dominant inclusion-body, 603902 (3)	HBB	141900	11p15.5	Hematological
1486	Thalassemia, delta- (3)	HBD	142000	11p15.5	Hematological
1486	Thalassemia due to Hb Lepore (3)	HBD	142000	11p15.5	Hematological
1486	Thalassemia, Hispanic gamma-delta-beta (3)	LCRB	152424	11p15.5	Hematological
1486	Thalassemias, alpha- (3)	HBA1	141800	16pter-p13.3	Hematological
1486	Thalassemias, beta- (3)	HBB	141900	11p15.5	Hematological
1490	Thanatophoric dysplasia, types I and II, 187600 (3)	FGFR3, ACH	134934	4p16.3	Skeletal
1491	Thiamine-responsive megaloblastic anemia syndrome, 249270 (3)	SLC19A2, THTR1	603941	1q23.3	Hematological
1493	Thrombocythemia, essential, 187950 (3)	JAK2	147796	9p24	Hematological
1493	Thrombocythemia, essential, 187950 (3)	THPO, MGDF, MPLLG, TPO	600044	3q26.3-q27	Hematological
1494	Thrombocytopenia-2, 188000 (3)	FLJ14813, THC2	608221	10p12.1	Hematological
1494	Thrombocytopenia, congenital amegakaryocytic, 604498 (3)	MPL, TPOR, MPLV	159530	1p34	Hematological
1494	Thrombocytopenia, X-linked, 313900 (3)	WAS, IMD2, THC	300392	Xp11.23-p11.22	Hematological
1494	Thrombocytopenia, X-linked, intermittent, 313900 (3)	WAS, IMD2, THC	300392	Xp11.23-p11.22	Hematological
1494	Thromboembolism susceptibility due to factor V Leiden (3)	F5	227400	1q23	Hematological
1497	Thrombophilia due to factor V Liverpool (3)	F5	227400	1q23	Hematological
1497	Thrombophilia due to heparin cofactor II deficiency (3)	HCF2, HC2, SERPIND1	142360	22q11	Hematological
1497	Thrombophilia due to HRG deficiency (3)	HRG	142640	3q27	Hematological
1497	Thrombophilia due to protein C deficiency (3)	PROC	176860	2q13-q14	Hematological
1497	Thrombophilia due to thrombomodulin defect (3)	THBD, THRM	188040	20p11.2	Hematological
1497	Thrombophilia, dysfibrinogenemic (3)	FGB	134830	4q28	Hematological
1497	Thrombophilia, dysfibrinogenemic (3)	FGG	134850	4q28	Hematological
1497	Thrombosis, hyperhomocysteinemic (3)	CBS	236200	21q22.3	Hematological
1497	Thrombotic thrombocytopenic purpura, familial, 274150 (3)	ADAMTS13, VWFCP, TTP	604134	9q34	Hematological
1494	Thrombycytosis, susceptibility to, 187950 (3)	MPL, TPOR, MPLV	159530	1p34	Hematological
1502	Thymine-uraciluria (3)	DPYD, DPD	274270	1p22	Metabolic
1503	Thyroid adenoma, hyperfunctioning (3)	TSHR	603372	14q31	Cancer
1503	Thyroid carcinoma (3)	TP53, P53, LFS1	191170	17p13.1	Cancer
1503	Thyroid carcinoma, follicular, 188470 (3)	MINPP1, HIPER1	605391	10q23	Cancer
1503	Thyroid carcinoma, follicular, 188470 (3)	PTEN, MMAC1	601728	10q23.31	Cancer
1503	Thyroid carcinoma, follicular, somatic, 188470 (3)	HRAS	190020	11p15.5	Cancer
1503	Thyroid carcinoma, papillary, 188550 (3)	GOLGA5, RFG5, PTC5	606918	14q	Cancer
1503	Thyroid carcinoma, papillary, 188550 (3)	NCOA4, ELE1, PTC3	601984	10q11.2	Cancer
1503	Thyroid carcinoma, papillary, 188550 (3)	PCM1, PTC4	600299	8p22-p21.3	Cancer
1503	Thyroid carcinoma, papillary, 188550 (3)	PRKAR1A, TSE1, CNC1, CAR	188830	17q23-q24	Cancer
1503	Thyroid carcinoma, papillary, 188550 (3)	TIF1G, RFG7, PTC7	605769	1p13	Cancer
1503	Thyroid carcinoma, papillary, 188550 (3)	TRIM24, TIF1, TIF1A, PTC6	603406	7q32-q34	Cancer
3503	Thyroid hormone organification defect IIA, 274500 (3)	TPO, TPX	606765	2p25	Endocrine
3503	Thyroid hormone resistance, 188570 (3)	THRB, ERBA2, THR1	190160	3p24.3	Endocrine
3503	Thyroid hormone resistance, autosomal recessive, 274300 (3)	THRB, ERBA2, THR1	190160	3p24.3	Endocrine
1504	Thyrotoxic periodic paralysis, susceptibility to, 188580 (3)	CACNA1S, CACNL1A3, CCHL1A3	114208	1q32	Endocrine
1505	Thyrotropin-releasing hormone resistance, generalized (3)	TRHR	188545	8q23	Endocrine
1506	Thyroxine-binding globulin deficiency (3)	TBG	314200	Xq22.2	Hematological
1508	Tietz syndrome, 103500 (3)	MITF, WS2A	156845	3p14.1-p12.3	multiple
1509	Timothy syndrome, 601005 (3)	CACNA1C, CACNL1A1, CCHL1A1, TS	114205	12p13.3	multiple
1510	Toenail dystrophy, isolated, 607523 (3)	COL7A1	120120	3p21.3	Dermatological

1511	Tolbutamide poor metabolizer (3)	CYP2C9	601130	10q24	Unclassified
3512	Total iodide organification defect, 274500 (3)	TPO, TPX	606765	2p25	Endocrine
1514	Townes-Brocks branchiootorenal-like syndrome, 107480 (3)	SALL1, HSAL1, TBS	602218	16q12.1	multiple
1514	Townes-Brocks syndrome, 107480 (3)	SALL1, HSAL1, TBS	602218	16q12.1	multiple
1515	Transaldolase deficiency, 606003 (3)	TALDO1	602063	11p15.5-p15.4	Metabolic
1516	Transcobalamin II deficiency (3)	TCN2, TC2	275350	22q11.2-qter	Hematological
1518	Transient bullous of the newborn, 131705 (3)	COL7A1	120120	3p21.3	Dermatological
1519	Transposition of great arteries, dextro-looped, 217095 (3)	CFC1, CRYPTIC, HTX2	605194	2q21.1	Developmental
1519	Transposition of the great arteries, dextro-looped, 608808 (3)	THRAP2, PROSIT240, TRAP240L, KIAA1025	608771	12q24	Developmental
1520	Treacher Collins mandibulofacial dysostosis, 154500 (3)	TCOF1, MFD1	606847	5q32-q33.1	Developmental
1521	Tremor, familial essential, 2, 602134 (3)	HS1BP3, FLJ14249, ETM2	609359	2p24.1	Neurological
1522	Trichodontoosseous syndrome, 190320 (3)	DLX3, TDO	600525	17q21.3-q22	multiple
1524	Trichorhinophalangeal syndrome, type I, 190350 (3)	TRPS1	604386	8q24.12	Developmental
1524	Trichorhinophalangeal syndrome, type III, 190351 (3)	TRPS1	604386	8q24.12	Developmental
1525	Trichothiodystrophy (3)	ERCC3, XPB	133510	2q21	Dermatological
1525	Trichothiodystrophy, 601675 (3)	ERCC2, EM9	126340	19q13.2-q13.3	Dermatological
1525	Trichothiodystrophy, complementation group A, 601675 (3)	TGF2H5, TTDA, TFB5, C6orf175	608780	6p25.3	Dermatological
1525	Trichothiodystrophy, nonphotosensitive 1, 234050 (3)	TTDN1, C7orf11, ABHS	609188	7p14	Dermatological
1526	Trifunctional protein deficiency, type 1 (3)	HADHA, MTPA	600890	2p23	Metabolic
1526	Trifunctional protein deficiency, type II (3)	HADHB	143450	2p23	Metabolic
1528	Trismus-pseudocomptodactyly syndrome, 158300 (3)	MYH8	160741	17p13.1	multiple
1529	Tropical calcific pancreatitis, 608189 (3)	SPINK1, PSTI, PCTT, TATI	167790	5q32	Gastrointestinal
1530	Troyer syndrome, 275900 (3)	SPG20	607111	13q12.3	Neurological
1534	TSC2 angiomyolipomas, renal, modifier of, 191100 (3)	IFNG	147570	12q14	multiple
1533	Tuberculosis, susceptibility to (3)	IFNGR1	107470	6q23-q24	Respiratory
1533	Tuberculosis, susceptibility to, 607948 (3)	IFNG	147570	12q14	Respiratory
1534	Tuberous sclerosis-1, 191100 (3)	TSC1, LAM	605284	9q34	multiple
1534	Tuberous sclerosis-2, 191100 (3)	TSC2, LAM	191092	16p13.3	multiple
1536	Turcot syndrome, 276300 (3)	APC, GS, FPC	175100	5q21-q22	Cancer
1536	Turcot syndrome with glioblastoma, 276300 (3)	MLH1, COCA2, HNPCC2	120436	3p21.3	Cancer
1536	Turcot syndrome with glioblastoma, 276300 (3)	PMS2, PMSL2, HNPCC4	600259	7p22	Cancer
1538	Twinning, dizygotic, 276400 (3)	FSHR, ODG1	136435	2p21-p16	Unclassified
1540	Tyrosinemia, type I (3)	FAH	276700	15q23-q25	Metabolic
1540	Tyrosinemia, type II (3)	TAT	276600	16q22.1-q22.3	Metabolic
1540	Tyrosinemia, type III (3)	HPD	276710	12q24-qter	Metabolic
1542	Ullrich congenital muscular dystrophy, 254090 (3)	COL6A1, OPLL	120220	21q22.3	Muscular
1542	Ullrich congenital muscular dystrophy, 254090 (3)	COL6A3	120250	2q37	Muscular
1542	Ullrich scleroatonic muscular dystrophy, 254090 (3)	COL6A2	120240	21q22.3	Muscular
1543	Ulnar-mammary syndrome, 181450 (3)	TBX3	601621	12q24.1	multiple
1544	Unipolar depression, susceptibility to, 608516 (3)	TPH2, NTPH	607478	12q21.1	Psychiatric
1545	Unna-Thost disease, nonepidermolytic, 600962 (3)	KRT1	139350	12q13	Dermatological
1550	Urolithiasis, 2,8-dihydroxyadenine (3)	APRT	102600	16q24.3	Metabolic
1550	Urolithiasis, hypophosphatemic (3)	SLC17A2, NPT2	182309	5q35	Metabolic
1551	Usher syndrome, type 1B (3)	MYO7A, USH1B, DFNB2, DFNA11	276903	11q13.5	multiple
1551	Usher syndrome, type 1C, 276904 (3)	USH1C, DFNB18	605242	11p15.1	multiple
1551	Usher syndrome, type 1D, 601067 (3)	CDH23, USH1D	605516	10q21-q22	multiple
1551	Usher syndrome, type 1F, 602083 (3)	PCDH15, DFNB23	605514	10q21-q22	multiple
1551	Usher syndrome, type 1G, 606943 (3)	SANS, USH1G	607696	17q24-q25	multiple
1551	Usher syndrome, type 2A, 276901 (3)	USH2A	608400	1q41	multiple

1551	Usher syndrome, type 3, 276902 (3)	USH3A, USH3	606397	3q21-q25	multiple
1551	Usher syndrome, type IIC, 605472 (3)	MASS1, VLGR1, KIAA0686, FEB4, USH2C	602851	5q14	multiple
1552	Uterine leiomyoma (3)	HMGA2, HMGIC, BABL, LIPO	600698	12q14.3	Cancer
1553	UV-induced skin damage, vulnerability to (3)	MC1R	155555	16q24.3	Dermatological
1554	van Buchem disease, type 2, 607636 (3)	LRP5, BMND1, LRP7, LR3, OPPG, VBCH2	603506	11q13.4	Unclassified
3554	van der Woude syndrome, 119300 (3)	IRF6, VWS, LPS, PIT, PPS, OFC6	607199	1q32-q41	Developmental
1555	VATER association with hydrocephalus, 276950 (3)	PTEN, MMAC1	601728	10q23.31	multiple
1556	Velocardiofacial syndrome, 192430 (3)	TBX1, DGS, CTHM, CAFS, TGA, DORV, VCFS, DGCR	602054	22q11.2	multiple
1557	Venous malformations, multiple cutaneous and mucosal, 600195 (3)	TEK, TIE2, VMCM	600221	9p21	Cardiovascular
1557	Venous thrombosis, susceptibility to (3)	SERPINA10, ZPI	605271	14q32.1	Cardiovascular
3558	Ventricular fibrillation, idiopathic, 603829 (3)	SCN5A, LQT3, IVF, HB1, SSS1	600163	3p21	Cardiovascular
1558	Ventricular tachycardia, idiopathic, 192605 (3)	GNAI2, GNAI2B, GIP	139360	3p21	Cardiovascular
1558	Ventricular tachycardia, stress-induced polymorphic, 604772 (3)	CASQ2	114251	1p13.3-p11	Cardiovascular
1558	Ventricular tachycardia, stress-induced polymorphic, 604772 (3)	RYR2, VTSIP	180902	1q42.1-q43	Cardiovascular
1559	Vertical talus, congenital, 192950 (3)	HOXD10, HOX4D	142984	2q31-q32	Skeletal
1562	Viral infections, recurrent (3)	FCGR3A, CD16, IGFR3	146740	1q23	Immunological
1562	Viral infection, susceptibility to (3)	OAS1, OIAS	164350	12q24.2	Immunological
1563	Virilization, maternal and fetal, from placental aromatase deficiency (3)	CYP19A1, CYP19, ARO	107910	15q21.1	Developmental
1565	Vitamin K-dependent clotting factors, combined deficiency of, 2, 607473 (3)	VKORC1, VKOR, VKCFD2, FLJ00289	608547	16p11.2	Hematological
1565	Vitamin K-dependent coagulation defect, 277450 (3)	GGCX	137167	2p12	Hematological
1566	Vitelliform macular dystrophy, adult-onset, 608161 (3)	VMD2	607854	11q13	Ophthamological
1570	VLCAD deficiency, 201475 (3)	ACADVL, VLCAD	609575	17p13	Metabolic
1571	Vohwinkel syndrome, 124500 (3)	GJB2, CX26, DFNB1, PPK, DFNA3, KID, HID	121011	13q11-q12	multiple
1571	Vohwinkel syndrome with ichthyosis, 604117 (3)	LOR	152445	1q21	multiple
1572	von Hippel-Lindau disease, modification of, 193300 (3)	CCND1, PRAD1, BCL1	168461	11q13	Cancer
1572	von Hippel-Lindau syndrome, 193300 (3)	VHL	608537	3p26-p25	Cancer
3572	von Willebrand disease (3)	VWF, F8VWF	193400	12p13.3	Hematological
1573	Waardenburg-Shah syndrome, 277580 (3)	EDNRB, HSCR2, ABCDS	131244	13q22	multiple
1573	Waardenburg-Shah syndrome, 277580 (3)	SOX10, WS4	602229	22q13	multiple
1574	Waardenburg syndrome/albinism, digenic, 103470 (3)	TYR	606933	11q14-q21	multiple
1574	Waardenburg syndrome/ocular albinism, digenic, 103470 (3)	MITF, WS2A	156845	3p14.1-p12.3	multiple
1574	Waardenburg syndrome, type I, 193500 (3)	PAX3, WS1, HUP2, CDHS	606597	2q35	multiple
1574	Waardenburg syndrome, type IIA, 193510 (3)	MITF, WS2A	156845	3p14.1-p12.3	multiple
1574	Waardenburg syndrome, type III, 148820 (3)	PAX3, WS1, HUP2, CDHS	606597	2q35	multiple
1574	Waardenburg syndrome, typ IID, 608890 (3)	SNAI2, SLUG, WS2D	602150	8q11	multiple
1575	Wagner syndrome, 143200 (3)	COL2A1	120140	12q13.11-q13.2	Ophthamological
1576	WAGR syndrome, 194072 (3)	WT1	607102	11p13	multiple
1578	Walker-Warburg syndrome, 236670 (3)	FCMD	607440	9q31	multiple
1578	Walker-Warburg syndrome, 236670 (3)	POMT1	607423	9q34.1	multiple
1578	Warburg micro syndrome 1, 600118 (3)	RAB3GAP, WARBM1, P130	602536	2q21.3	multiple
1580	Warfarin resistance, 122700 (3)	VKORC1, VKOR, VKCFD2, FLJ00289	608547	16p11.2	Hematological
1580	Warfarin sensitivity, 122700 (3)	CYP2C9	601130	10q24	Hematological
1580	Warfarin sensitivity (3)	F9, HEMB	306900	Xq27.1-q27.2	Hematological
1581	Watson syndrome, 193520 (3)	NF1, VRNF, WSS, NFNS	162200	17q11.2	Cancer
1582	Weaver syndrome, 277590 (3)	NSD1, ARA267, STO	606681	5q35	Developmental
1583	Wegener-like granulomatosis (3)	TAP2, ABCB3, PSF2, RING11	170261	6p21.3	Immunological

1585	Weill-Marchesani syndrome, dominant, 608328 (3)	FBN1, MFS1, WMS	134797	15q21.1	Connective tissue
1585	Weill-Marchesani syndrome, recessive, 277600 (3)	ADAMTS10, WMS	608990	19p13.3-p13.2	Connective tissue
1586	Weissenbacher-Zweymuller syndrome, 277610 (3)	COL11A2, STL3, DFNA13	120290	6p21.3	Connective tissue
1588	Werner syndrome, 277700 (3)	RECQL2, RECQ3, WRN	604611	8p12-p11.2	multiple
1589	Wernicke-Korsakoff syndrome, susceptibility to, 277730 (3)	TKT	606781	3p14.3	Metabolic
1590	Weyers acrodental dysostosis, 193530 (3)	EVC	604831	4p16	Skeletal
1591	WHIM syndrome, 193670 (3)	CXCR4, D2S201E, NPY3R, WHIM	162643	2q21	Immunological
1592	White sponge nevus, 193900 (3)	KRT13	148065	17q21-q22	Connective tissue
1592	White sponge nevus, 193900 (3)	KRT4, CYK4	123940	12q13	Connective tissue
1594	Williams-Beuren syndrome, 194050 (3)	ELN	130160	7q11.2	multiple
1595	Wilms tumor, 194070 (3)	BRCA2, FANCD1	600185	13q12.3	Cancer
1595	Wilms tumor, somatic, 194070 (3)	GPC3, SDYS, SGBS1	300037	Xq26	Cancer
1595	Wilms tumor susceptibility-5, 601583 (3)	POU6F2, WTSL, WT5	609062	7p14-p13	Cancer
1595	Wilms tumor, type 1, 194070 (3)	WT1	607102	11p13	Cancer
1596	Wilson disease, 277900 (3)	ATP7B, WND	606882	13q14.3-q21.1	Metabolic
1598	Wiskott-Aldrich syndrome, 301000 (3)	WAS, IMD2, THC	300392	Xp11.23-p11.22	Immunological
1599	Witkop syndrome, 189500 (3)	MSX1, HOX7, HYD1, OFC5	142983	4p16.1	Developmental
1600	Wolcott-Rallison syndrome, 226980 (3)	EIF2AK3, PEK, PERK, WRS	604032	2p12	Bone
1601	Wolff-Parkinson-White syndrome, 194200 (3)	PRKAG2, WPWS	602743	7q36	Cardiovascular
1603	Wolfram syndrome, 222300 (3)	WFS1, WFRS, WFS, DFNA6	606201	4p16.1	Metabolic
1604	Wolman disease (3)	LIPA	278000	10q24-q25	Metabolic
1607	Xanthinuria, type I, 278300 (3)	XDH	607633	2p23-p22	Metabolic
1608	Xeroderma pigmentosum, group A (3)	XPA	278700	9q22.3	Dermatological
1608	Xeroderma pigmentosum, group B (3)	ERCC3, XPB	133510	2q21	Dermatological
1608	Xeroderma pigmentosum, group C (3)	XPC, XPCC	278720	3p25	Dermatological
1608	Xeroderma pigmentosum, group D, 278730 (3)	ERCC2, EM9	126340	19q13.2-q13.3	Dermatological
1608	Xeroderma pigmentosum, group E, DDB-negative subtype, 278740 (3)	DDB2	600811	11p12-p11	Dermatological
1608	Xeroderma pigmentosum, group F, 278760 (3)	ERCC4, XPF	133520	16p13.3-p13.13	Dermatological
1608	Xeroderma pigmentosum, group G, 278780 (3)	ERCC5, XPG	133530	13q33	Dermatological
1608	Xeroderma pigmentosum, variant type, 278750 (3)	POLH, XPV	603968	6p21.1-p12	Dermatological
1610	X-inactivation, familial skewed, 300087 (3)	XIC, XCE, XIST, SXI1	314670	Xq13.2	Unclassified
1611	XLA and isolated growth hormone deficiency, 307200 (3)	BTK, AGMX1, IMD1, XLA, AT	300300	Xq21.3-q22	Immunological
1613	Yellow nail syndrome, 153300 (3)	FOXC2, FKHL14, MFH1	602402	16q24.3	Immunological
1614	Yemenite deaf-blind hypopigmentation syndrome, 601706 (3)	SOX10, WS4	602229	22q13	multiple
1615	Zellweger syndrome-1, 214100 (3)	PEX1, ZWS1	602136	7q21-q22	multiple
1615	Zellweger syndrome, 214100 (3)	PEX10, NALD	602859	Chr.1	multiple
1615	Zellweger syndrome, 214100 (3)	PEX13, ZWS, NALD	601789	2p15	multiple
1615	Zellweger syndrome, 214100 (3)	PEX14	601791	1p36.2	multiple
1615	Zellweger syndrome, 214100 (3)	PEX26	608666	22q11.21	multiple
1615	Zellweger syndrome, 214100 (3)	PXF, HK33, D1S2223E, PEX19	600279	1q22	multiple
1615	Zellweger syndrome, 214100 (3)	PXR1, PEX5, PTS1R	600414	12p13.3	multiple
1615	Zellweger syndrome-2 (3)	ABCD3, PXMP1, PMP70	170995	1p22-p21	multiple
1615	Zellweger syndrome-3 (3)	PXMP3, PAF1, PMP35, PEX2	170993	8q21.1	multiple
1615	Zellweger syndrome, complementation group 9 (3)	PEX16	603360	11p12-p11.2	multiple
1615	Zellweger syndrome, complementation group G, 214100 (3)	PEX3	603164	6q23-q24	multiple
1617	Zlotogora-Ogur syndrome, 225000 (3)	HVEC, PVRL1, PVRR1, PRR1	600644	11q23-q24	multiple