REVIEW ARTICLE

The morbid anatomy of the human genome: chromosomal location of mutations causing disease

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Abstract

Information is given in tabular form derived from a synopsis of the human gene map which has been updated continuously since 1973 as part of *Mendelian Inheritance in Man* (Johns Hopkins University Press, 10th ed, 1992) and of OMIM (Online *Mendelian Inheritance in Man*, available generally since 1987). The part of the synopsis reproduced here consists of chromosome by chromosome gene lists of loci for which there are associated disorders (table 1), a pictorial representation of this information (fig 1a–d), and an index of disorders for which the causative mutations have been mapped (table 2).

In table 1, information on genes that have been located to specific chromosomal positions and are also the site of disease producing mutations is arranged by chromosome, starting with chromosome 1 and with the end of the short arm of the chromosome in each case.

In table 2 an alphabetised list of these disorders and the chromosomal location of the mutation in each case are provided. Both in the 'Disorder' field of table 1 and in table 2, the numbers 1, 2, or 3 in parentheses after the name of the disorder indicate that its chromosomal location was determined by mapping of the wildtype gene (1), by mapping of the clinical phenotype (2), or by both strategies (3).

(*J Med Genet* 1993;**30**:1-26)

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Received 12 October 1992.

The fields in the listing by chromosome (table 1) are as follows:

Location – that is, chromosomal site (p = short arm; q = long arm; numbers = band; ter = end).

Symbol – The symbol approved by the Nomenclature Committee of the Human Gene Mapping Workshops and their successor organisation is given first; alternative symbols are included.

Status – C = confirmed; P = provisional; L = 'in limbo' (that is, tentative or inconsistent). The 'in limbo' entries are not listed in fig 1.

Title – name of gene locus.

MIM# – This is the number in McKusick's *Mendelian Inheritance in Man*, 10th ed (1992) and its continuously updated online version OMIM. (For historical reasons, the number may sometimes indicate location of the entry in the 'dominant catalogue' because the wildtype gene was characterised and mapped before the recessive disorder resulting from mutation at that site. The practice is to create only one entry in OMIM for each gene locus.)

Method of mapping

A = in situ DNA-RNA or DNA-DNA annealing ('hybridisation'), for example, ribosomal RNA genes to acrocentric chromosomes; kappa light chain genes to chromosome 2.

AAS = deductions from the amino acid sequence of proteins, for example, linkage of delta and beta globin loci from study of hemoglobin Lepore.

C = chromosome mediated gene transfer (CMGT), for example, cotransfer of galactokinase and thymidine kinase.

Ch = chromosomal change associated with particular phenotype and not proved to represent linkage (Fc), deletion (D), or virus effect (V), for example, loss of 13q14 band in some cases of retinoblastoma.

D = deletion or dosage mapping (concurrence of chromosomal deletion and phenotypic evidence of hemizygosity), trisomy mapping (presence of three alleles in the case of a highly polymorphic locus), or gene dosage effects (correlation of trisomic state of part or all of a chromosome with 50% more gene product). Includes 'loss of heterozygosity' (loss of alleles) in malignancies. Examples: acid phosphatase-1 to chromosome 2; glutathione reductase to chromosome 8. Includes DNA dosage, for example, fibrinogen loci to 4q2. Dosage mapping also includes coamplification in tumour cells.

EM = exclusion mapping, that is, narrowing the possible location of loci by exclusion of parts of the map by deletion mapping, extended to include negative lod scores from families with marker chromosomes and negative lod scores with other assigned loci, for example, support for assignment of MNSs to 4q.

F = linkage study in families, for example, linkage of ABO blood group and nail-patella syndrome. (When a chromosomal heteromorphism or rearrangement is one trait, Fc is used, for example, Duffy blood group locus on chromosome 1. When one or both of the linked loci are identified by a DNA polymorphism, Fd is used, for example, Huntington's disease on chromosome 4. F = L in the HGM workshops.)

H = based on presumed homology, for example, proposed assignment of TF to 3q. Mainly heuristic or confirmatory.

HS = DNA/cDNA molecular hybridisation in solution ('Cot analysis').

L = lyonisation, for example, OTC to X chromosome. (L = symbol for family linkage study in the HGM workshops.)

LD = linkage disequilibrium, for example, beta and delta globin genes.

M = Microcell mediated gene transfer (MMGT), for example, a collagen gene (COL1A1) to chromosome 17.

OT = ovarian teratoma (centromere mapping), for example, PGM3 and centromere of chromosome 6.

Pcm = PCR of microdissected chromosome segments (see REl).

Psh = PCR of somatic cell hybrid DNA.

R = radiation hybrids, that is, irradiation of cells followed by 'rescue' through fusion with non-irradiated (non-human) cells (Goss-Harris method of radiation-induced gene segregation), for example, order of genes on Xq.

RE = restriction endonuclease techniques, for example, fine structure map of the beta globin cluster (HBBC) on 11p; physical linkage of 3 fibrinogen genes (on 4q).

REa = combined with somatic cell hybridisation, for example, NAG (HBBC) to 11p.

REb = combined with chromosome sorting, for example, insulin to 11p. Includes Lebo's adaptation (dual laser chromosome sorting and spot blot DNA analysis), for example, MGP to 11q. (For this method, using flow sorted chromosomes, W was the symbol adopted by the HGM workshops.)

REc = hybridisation of cDNA to genomic fragment (by YAC, PFGE, microdissection, etc), for example, A-11 on Xq.

REl = isolation of gene from chromosome specific genomic library (see Pcm).

REn = neighbour analysis in restriction fragments, for example, in pulsed field gel electrophoresis (PFGE).

S = 'segregation' (cosegregation) of human cellular traits and human chromosomes (or segments of chromosomes) in particular clones from interspecies somatic cell hybrids, for example, thymidine kinase to chromosome 17. When with restriction enzyme, REa; with hybridisation in solution, HS.

V = induction of microscopically evident chromosomal change by a virus, for example, adenovirus 12 changes on chromosomes 1 and 17.

X/A = X-autosome translocation in female with X linked recessive disorder, for example, assignment of Duchenne muscular dystrophy to Xp21.

Disorder – Allelic disorders are separated by semicolons. Brackets [] indicate a 'non-disease', that is, a variation with no definite adverse consequences. Braces { } indicate specific susceptibility or resistance with monogenic basis. (1) = wildtype gene mapped. (2) = disease phenotype mapped. (3) = both wildtype gene and disease phenotype mapped.

Mouse – Mouse chromosome carrying homologous gene.

References for the mapping information and additional information about the loci and disorders are provided in *Mendelian Inheritance in Man*, 10th ed (McKusick, 1992) and OMIM™ (Online *Mendelian Inheritance in Man*). To obtain information on accessing OMIM, contact:

In the United States: GDB/OMIM User Support, Welch Medical Library, 1830 E Monument Street, 3rd Floor, Baltimore, MD 21205, USA (Telephone: 410-955-7058, Fax: 410-955-0054, e-mail: help@welch.jhu.edu).

In the United Kingdom: Christine Bates, Human Gene Mapping Program Resource Centre, CRC, Watford Road, Harrow, Middx HA1 3UJ, UK (Telephone: 44-81-869-3446, Fax: 44-81-869-3807, e-mail: cbates@uk.ac.crc).

In Germany: Otto Ritter, German Cancer Research Centre (DKFZ), Molecular Biophysics Group, Im Neuenheimer Feld 280, D-6900 Heidelberg 1, FRG (Telephone: 49-6221-42-2372, Fax: 49-6221-40-1271, e-mail: dok261@cvx12.dkfz-heidelberg.de).

In Australia: Alex Reisner, ANGIS, Electrical Engineering Building, JO3, University of Sydney, Sydney, NSW 2006, Australia, (Telephone: 61–2–692–294, Fax 61–2–692–3847, e-mail: reisner@ee.su.oz.au).

Discussion

In these listings, 738 disorders in total are indicated as assigned to specific chromosomes and, in most instances, to specific regions of those chromosomes. (More than 50 other disorders are known to be caused by mutations in genes on the X chromosome but their regional location is not known and therefore they have not been included here.) The 738 disorders are distributed over 625 loci; many loci have more than one allelic mutation producing distinct phenotypes.

Some of the disorders (labelled with the number 1) have been mapped to specific sites by virtue of mapping of the wildtype gene; many forms of chronic non-spherocytic haemolytic anaemia such as those due to pyruvate kinase deficiency, triosephosphate isomerase deficiency, and bisphosphoglycerate mutase deficiency are examples.

Other disorders (labelled with the number 2) are mapped only on the basis of the clinical phenotype which is found to be linked to markers at a particular chromosomal site or the phenotype is found in association with a

chromosomal aberration. Huntington's disease is an example of a disorder mapped by linkage to DNA markers.

Yet other disorders (labelled with the number 3) have been mapped by both approaches. The form of elliptocytosis due to a defect in the gene encoding protein 4.1 is an early example, and piebaldism (due to mutation in the KIT gene) is a recent one.

Chromosomal aberrations have contributed to the mapping of Mendelian disorders to an extent that perhaps would not have been predicted. Both translocations and deletions have been informative. Because of the extensive homology of synteny shown between mouse and man, comparative mapping has also had great heuristic and confirmatory value in identifying the location of genes and genetic disorders in the human.

Mapping of genetic disorders has shed light on both allelism and non-allelism. Both phenomena are illustrated by the haemoglobinopathies and the collagenopathies. Because of the heteromeric structure of the haemoglobin molecule, thalassaemia, Heinz body haemolytic anaemia, erythraemia, or methaemoglobinaemia can result from mutation in either the α globin gene on chromosome 16 or the β globin gene on chromosome 11. Similarly, in the case of the type I collagen molecule, which is also heteromeric, two or more different forms of osteogenesis imperfecta, as well as the distinct disorder Ehlers-Danlos syndrome, can occur from various mutation in either the gene for the α1 chain (COL1A1) on chromosome 17 or that of the α2 chain (COL1A2) on chromosome 7.

Allelic series as the basis of phenotypic diversity are well illustrated by Duchenne and Becker muscular dystrophies and by the several forms of spinal muscular atrophy. Gardner syndrome and adenomatous polyposis coli (without extra-bowel manifestations) are demonstrably allelic; indeed the fact that the same mutation has been shown to cause one or the other of these two disorders in different members of the same family indicates that the two syndromes represent variability of expression.

Mapping information suggests that some syndromes with a combination of manifestations may result from mutation in a complex locus, that is, the particular pattern of components may be determined by the part of the gene affected by the mutation. Von Hippel-Lindau syndrome and multiple endocrine neoplasia type II are possible examples. Close linkage of mutant genes, each responsible for a specific component, in general is rejected as the cause of genetic syndromes, in favour of pleiotropism. The bona fide nature of some so called contiguous gene syndromes is unquestionable, however.

Somatic cell genetic disease is particularly well illustrated by many forms of neoplasia, both solid tumours and leukaemias, that have been traced to genes located at specific sites. Many are listed among the disorders here. Some congenital malformations also represent somatic cell genetic disorders. The Happle hypothesis holds that some disorders are due to the mosaic state of mutations which would be lethal if present in the non-mosaic state, even in the heterozygote. The McCune-Albright syndrome (Albright polyostotic fibrous dysplasia), pseudohypoparathyroidism (Albright hereditary osteodystrophy), and growth hormone secreting pituitary tumour comprise a remarkable set of genetic diseases illustrating an allelic series. The first, a somatic cell genetic disease, is due to an activating mutation in the GNAS1 gene occurring in early embryogenesis; the second is a loss of function mutation of the GNAS1 gene, and the third represents an activating somatic mutation occurring in a single cell of the anterior pituitary.

We are indebted to colleagues who have participated in the Human Gene Mapping Workshops 1973 to 1991 and assisted in the collation of data on the human gene map. The development and maintenance of *Mendelian Inheritance in Man* and its online version OMIM was supported in part by the Howard Hughes Medical Institute from 1986 to 1991. It is now supported, in the main, jointly by the NIH and the DOE as part of the Genome Data Base (GDB), the repository for mapping information coming from the Human Genome Project.

1 McKusick VA: *Mendelian inheritance in man*, 10th ed. Baltimore: The Johns Hopkins University Press, 1992.

Editorial note. It is our intention to publish an annual update of this information.

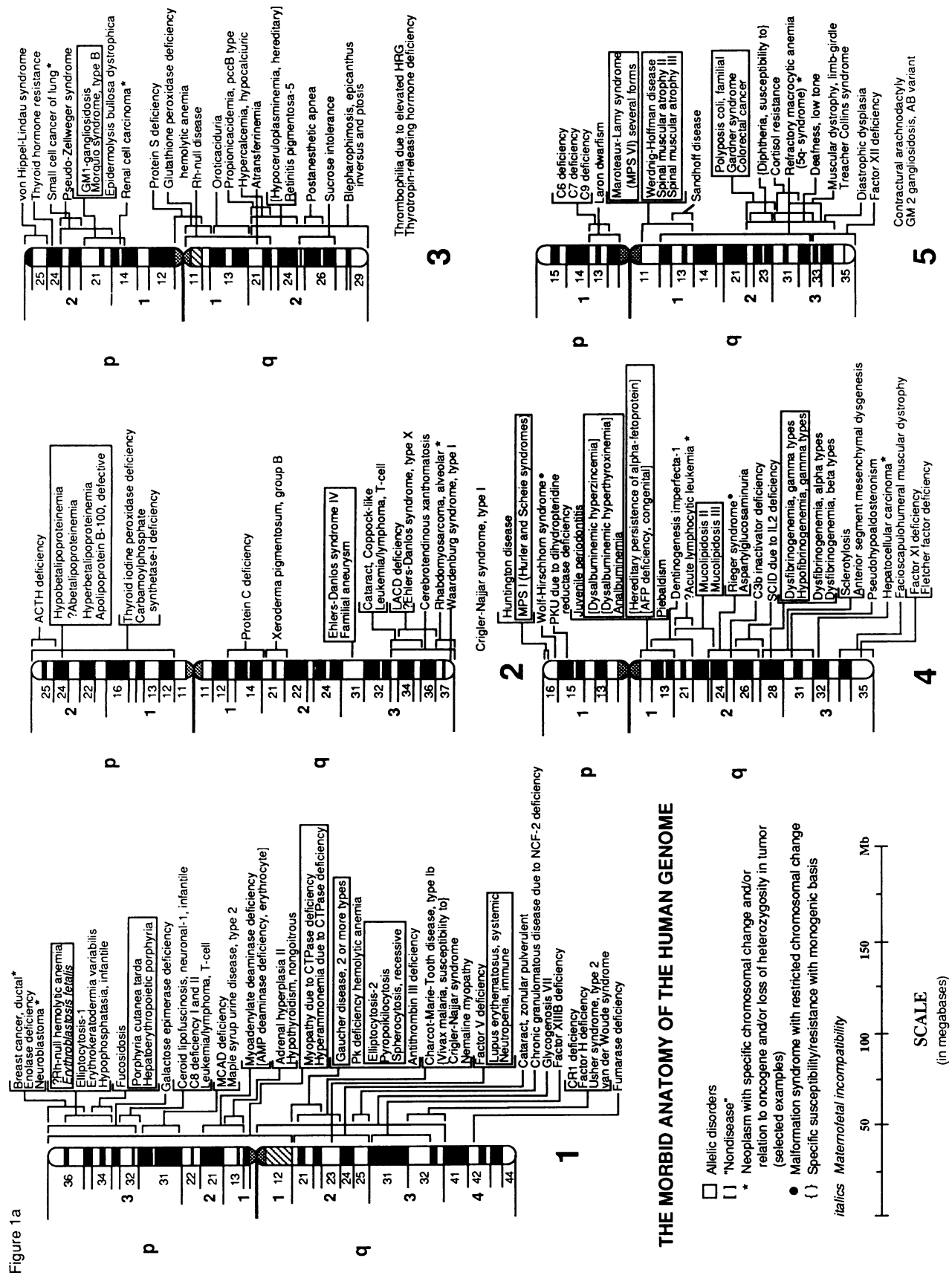
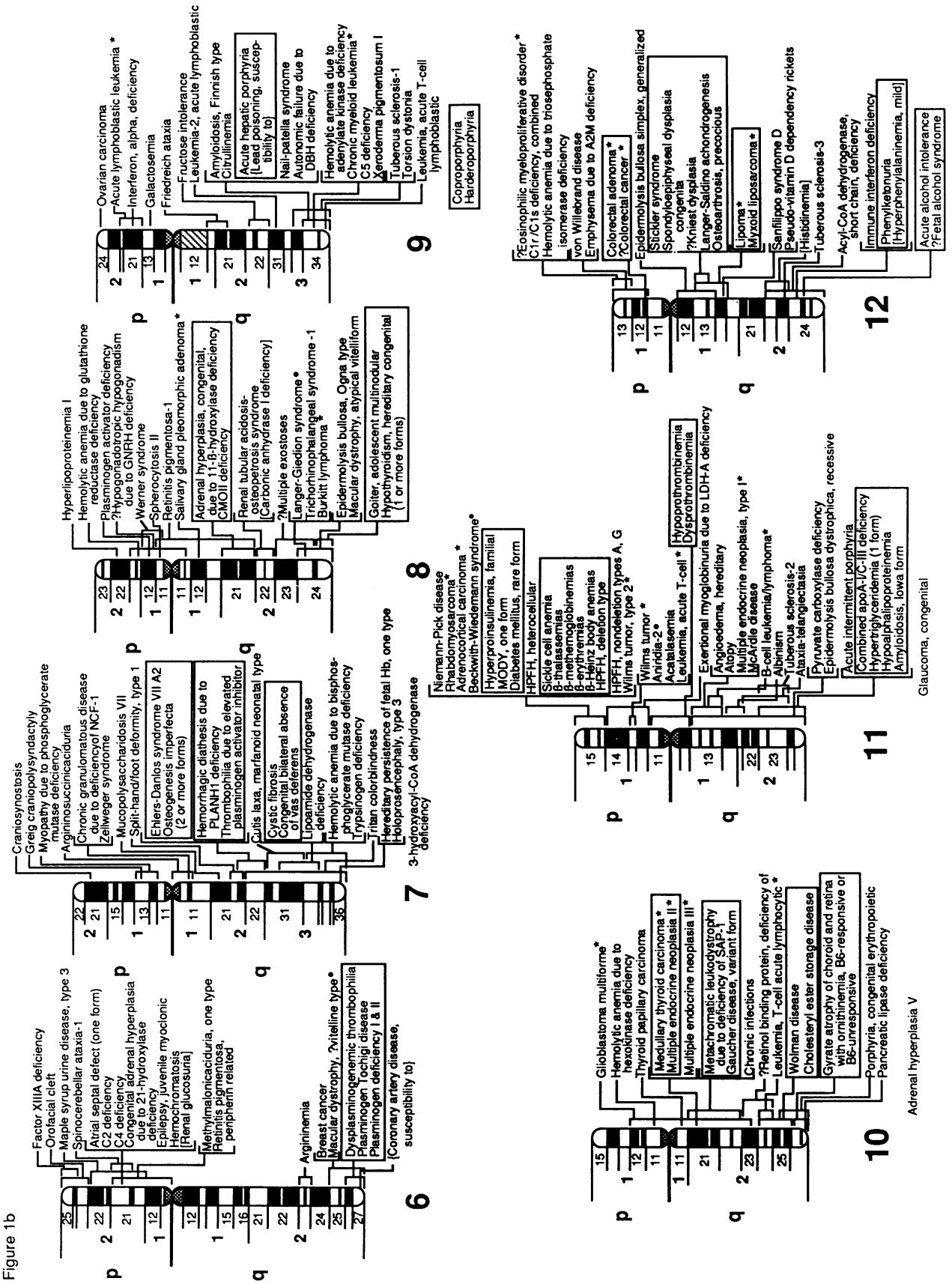
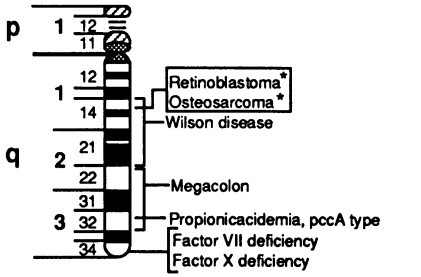
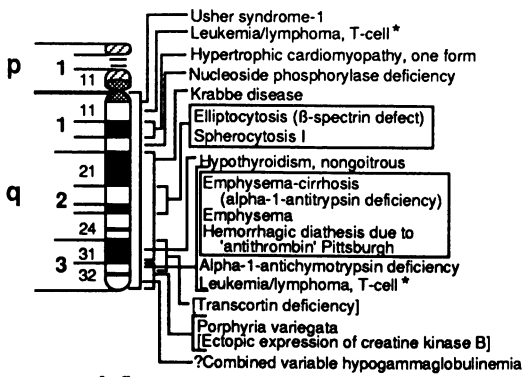


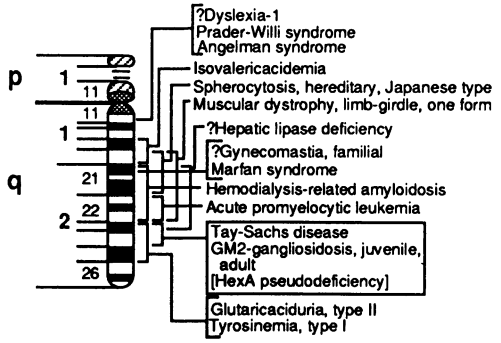
Figure 1a–d The morbid anatomy of the human genome: disorders with confirmed or provisional assignments have been included. Because of the large number of disorders assigned to specific regions of the X chromosome, only selected ones are represented here.

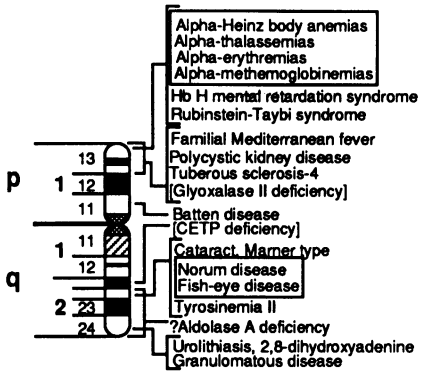
Figure 1b

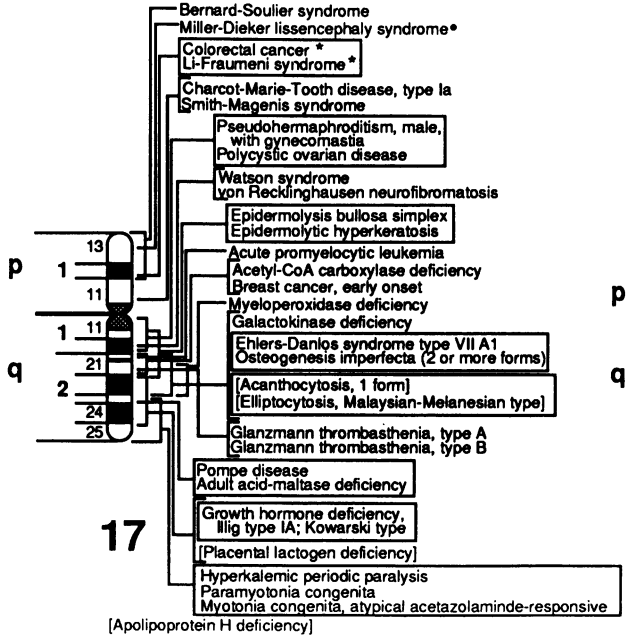

Factor XIIIA deficiency
Orofacial cleft
Maple syrup urine disease, type 3
Spinocerebellar ataxia-1
Atrial septal defect (one form)
C2 deficiency
C4 deficiency
Congenital adrenal hyperplasia due to 21-hydroxylase deficiency
Epilepsy, juvenile myoclonic
Hemochromatosis
[Renal glucosuria]
Methylmalonicaciduria, one type
Retinitis pigmentosa, peripherin related
Argininemia
Breast cancer
Macular dystrophy, ?vitelline type*
Dysplasminogenemic thrombophilia
Plasminogen Tochigi disease
Plasminogen deficiency I & II
[Coronary artery disease, susceptibility to]
6
Craniosynostosis
Greig craniopolysyndactyly
Myopathy due to phosphoglycerate mutase deficiency
Argininosuccinicaciduria
Chronic granulomatous disease due to deficiencyof NCF-1
Zellweger syndrome
Mucopolysaccharidosis VII
Split-hand/foot deformity, type 1
Ehlers-Danlos syndrome VII A2
Osteogenesis imperfecta
(2 or more forms)
Hemorrhagic diathesis due to PLANH1 deficiency
Thrombophilia due to elevated plasminogen activator inhibitor
Cutis laxa, marfanoid neonatal type
Cystic fibrosis
Congenital bilateral absence of vas deferens
Lipoamide dehydrogenase deficiency
Hemolytic anemia due to bisphosphoglycerate mutase deficiency
Trypsinogen deficiency
Tritan colorblindness
Hereditary persistence of fetal Hb, one type
Holoprosencephaly, type 3
3-hydroxyacyl-CoA dehydrogenase deficiency
7
Niemann-Pick disease
Rhabdomyosarcoma*
Adrenocortical carcinoma*
Beckwith-Wiedemann syndrome*
Hyperproinsulinemia, familial
MODY, one form
Diabetes mellitus, rare form
HPFH, heterocellular
Sickle cell anemia
β-thalassemias
β-methemoglobinemias
β-erythremias
β-Heinz body anemias
HPFH, deletion type
HPFH, nondeletion types A, G
Wilms tumor, type 2*
Wilms tumor *
Aniridia-2*
Acatalasemia
Leukemia, acute T-cell *
Hypoprothrombinemia
Dysprothrombinemia
8
Hyperlipoproteinemia I
Hemolytic anemia due to glutathione reductase deficiency
Plasminogen activator deficiency
?Hypogonadotropic hypogonadism due to GNRH deficiency
Werner syndrome
Spherocytosis II
Retinitis pigmentosa-1
Salivary gland pleomorphic adenoma*
Adrenal hyperplasia, congenital, due to 11-β-hydroxylase deficiency
CMOII deficiency
Renal tubular acidosis-osteopetrosis syndrome
[Carbonic anhydrase I deficiency]
?Multiple exostoses
Langer-Giedion syndrome•
Trichorhinophalangeal syndrome -1
Burkitt lymphoma*
Epidermolysis bullosa, Ogna type
Macular dystrophy, atypical vitelliform
Goiter, adolescent multinodular
Hypothyroidism, hereditary congenital (1 or more forms)
9
Ovarian carcinoma
Acute lymphoblastic leukemia *
Interferon, alpha, deficiency
Galactosemia
Friedreich ataxia
Fructose intolerance
Leukemia-2, acute lymphoblastic
Amyloidosis, Finnish type
Citrullinemia
Acute hepatic porphyria
[Lead poisoning, susceptibility to]
Nail-patella syndrome
Autonomic failure due to DBH deficiency
Hemolytic anemia due to adenylate kinase deficiency
Chronic myeloid leukemia*
C5 deficiency
Xeroderma pigmentosum I
Tuberous sclerosis-1
Torsion dystonia
Leukemia, acute T-cell lymphoblastic
Coproporphyria
Harderoporphyria
Glioblastoma multiforme*
Hemolytic anemia due to hexokinase deficiency
Thyroid papillary carcinoma
Medullary thyroid carcinoma*
Multiple endocrine neoplasia II*
Multiple endocrine neoplasia III*
Metachromatic leukodystrophy due to deficiency of SAP-1
Gaucher disease, variant form
Chronic infections
?Retinol binding protein, deficiency of
Leukemia, T-cell acute lymphocytic*
Wolman disease
Cholesteryl ester storage disease
Gyrate atrophy of choroid and retina with ornithinemia, B6-responsive or B6-unresponsive
Porphyria, congenital erythropoietic
Pancreatic lipase deficiency
10
Adrenal hyperplasia V
Exertional myoglobinuria due to LDH-A deficiency
Angioedema, hereditary
Atopy
Multiple endocrine neoplasia, type I*
McArdle disease
B-cell leukemia/lymphoma*
Albinism
Tuberous sclerosis-2
Ataxia-telangiectasia
Pyruvate carboxylase deficiency
Epidermolysis bullosa dystrophica, recessive
Acute intermittent porphyria
Combined apoA-I/C-III deficiency
Hypertriglyceridemia (1 form)
Hypoalphalipoproteinemia
Amyloidosis, Iowa form
Glaucoma, congenital
11
?Eosinophilic myeloproliferative disorder*
C1r /C1s deficiency, combined
Hemolytic anemia due to triosephosphate isomerase deficiency
von Willebrand disease
Emphysema due to A2M deficiency
Colorectal adenoma*
?Colorectal cancer *
Epidermolysis bullosa simplex, generalized
Stickler syndrome
Spondyloepiphyseal dysplasia congenita
?Kniest dysplasia
Langer-Saldino achondrogenesis
Osteoarthrosis, precocious
Lipoma*
Myxoid liposarcoma*
Sanfilippo syndrome D
Pseudo-vitamin D dependency rickets
[Histidinemia]
Tuberous sclerosis-3
Acyl-CoA dehydrogenase, short chain, deficiency
Immune interferon deficiency
Phenylketonuria
[Hyperphenylalaninemia, mild]
Acute alcohol intolerance
?Fetal alcohol syndrome
12

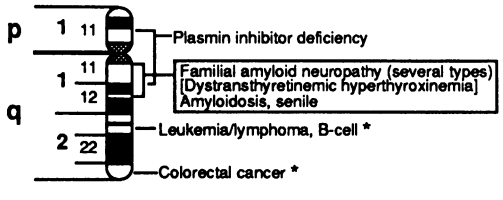

p
q
Retinoblastoma*
Osteosarcoma*
Wilson disease
Megacolon
Propionicacidemia, pccA type
Factor VII deficiency
Factor X deficiency
?Xeroderma pigmentosum (one type)
Breast cancer, ductal
13


Usher syndrome-1
Leukemia/lymphoma, T-cell *
Hypertrophic cardiomyopathy, one form
Nucleoside phosphorylase deficiency
Krabbe disease
Elliptocytosis (ß-spectrin defect)
Spherocytosis I
Hypothyroidism, nongoitrous
Emphysema-cirrhosis (alpha-1-antitrypsin deficiency)
Emphysema
Hemorrhagic diathesis due to 'antithrombin' Pittsburgh
Alpha-1-antichymotrypsin deficiency
Leukemia/lymphoma, T-cell *
Transcortin deficiency]
Porphyria variegata
[Ectopic expression of creatine kinase B]
?Combined variable hypogammaglobulinemia
14
Glycogen storage disease VI (Hers disease)


?Dyslexia-1
Prader-Willi syndrome
Angelman syndrome
Isovalericacidemia
Spherocytosis, hereditary, Japanese type
Muscular dystrophy, limb-girdle, one form
?Hepatic lipase deficiency
?Gynecomastia, familial
Marfan syndrome
Hemodialysis-related amyloidosis
Acute promyelocytic leukemia
Tay-Sachs disease
GM2-gangliosidosis, juvenile, adult
[HexA pseudodeficiency]
Glutaricaciduria, type II
Tyrosinemia, type I
Xeroderma pigmentosum, group F
15


Alpha-Heinz body anemias
Alpha-thalassemias
Alpha-erythremias
Alpha-methemoglobinemias
Hb H mental retardation syndrome
Rubinstein-Taybi syndrome
Familial Mediterranean fever
Polycystic kidney disease
Tuberous sclerosis-4
[Glyoxalase II deficiency]
Batten disease
[CETP deficiency]
Cataract, Marner type
Norum disease
Fish-eye disease
Tyrosinemia II
?Aldolase A deficiency
Urolithiasis, 2,8-dihydroxyadenine
Granulomatous disease
[Cystathioninuria]
Brody myopathy
16


Bernard-Soulier syndrome
Miller-Dieker lissencephaly syndrome•
Colorectal cancer *
Li-Fraumeni syndrome*
Charcot-Marie-Tooth disease, type Ia
Smith-Magenis syndrome
Pseudohermaphroditism, male, with gynecomastia
Polycystic ovarian disease
Watson syndrome
von Recklinghausen neurofibromatosis
Epidermolysis bullosa simplex
Epidermolytic hyperkeratosis
Acute promyelocytic leukemia
Acetyl-CoA carboxylase deficiency
Breast cancer, early onset
Myeloperoxidase deficiency
Galactokinase deficiency
Ehlers-Danlos syndrome type VII A1
Osteogenesis imperfecta (2 or more forms)
[Acanthocytosis, 1 form]
[Elliptocytosis, Malaysian-Melanesian type]
Glanzmann thrombasthenia, type A
Glanzmann thrombasthenia, type B
Pompe disease
Adult acid-maltase deficiency
Growth hormone deficiency, Illig type IA; Kowarski type
[Placental lactogen deficiency]
Hyperkalemic periodic paralysis
Paramyotonia congenita
Myotonia congenita, atypical acetazolaminde-responsive
[Apolipoprotein H deficiency]
17


Plasmin inhibitor deficiency
Familial amyloid neuropathy (several types)
[Dystransthyretinemic hyperthyroxinemia]
Amyloidosis, senile
Leukemia/lymphoma, B-cell *
Colorectal cancer *
Methemoglobinemia due to cytochrome b5 deficiency
18

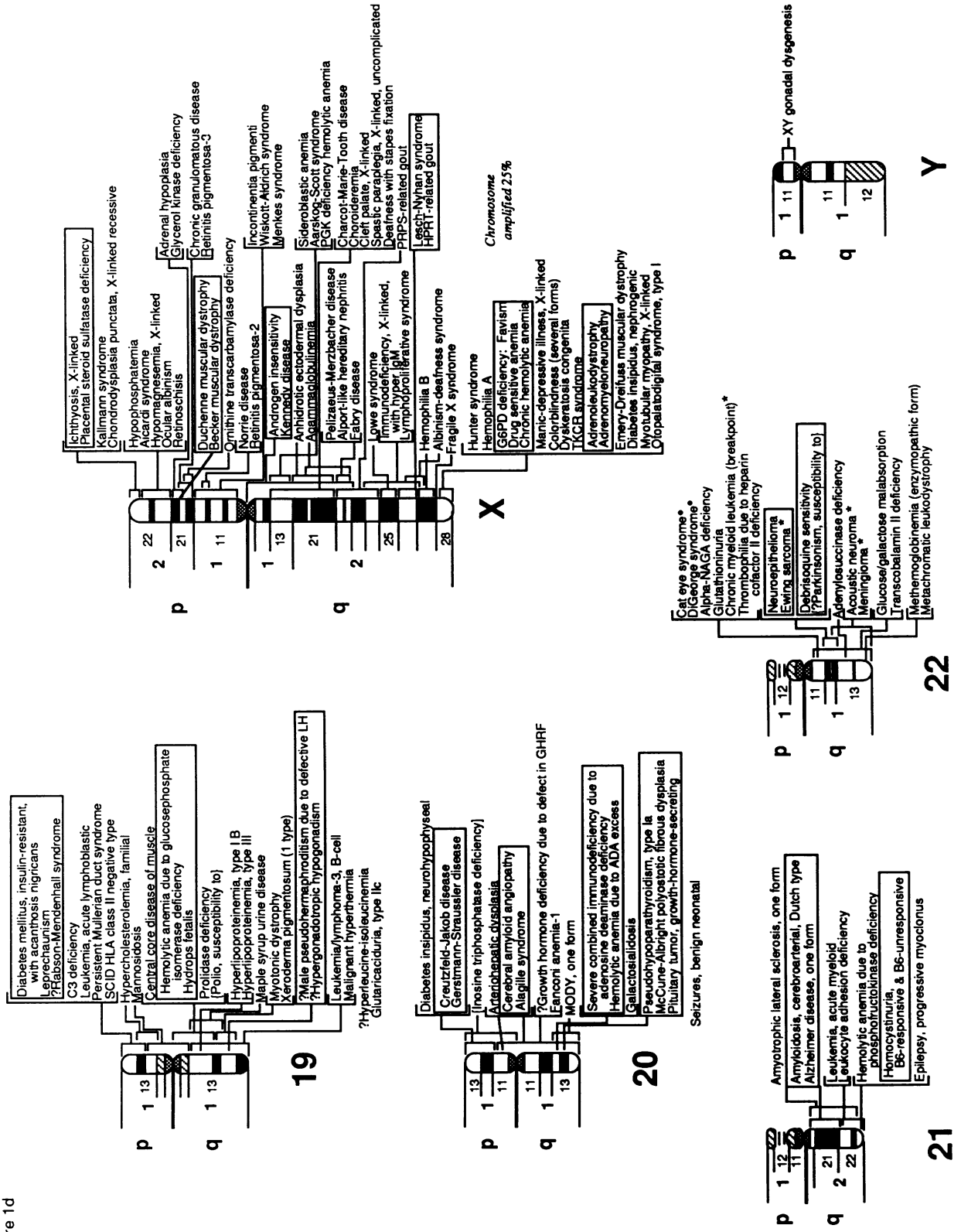

Ichthyosis, X-linked
Placental steroid sulfatase deficiency
Kallmann syndrome
Chondrodysplasia punctata, X-linked recessive
Hypophosphatemia
Aicardi syndrome
Oculo-cerebrorenal syndrome
Ocular albinism
Retinoschisis
Adrenal hypoplasia
Glycerol kinase deficiency
Chronic granulomatous disease
Retinitis pigmentosa-3
Duchenne muscular dystrophy
Becker muscular dystrophy
Ornithine transcarbamylase deficiency
Norrie disease
Retinitis pigmentosa-2
Androgen insensitivity
Kennedy disease
Anhidrotic ectodermal dysplasia
Agammaglobulinemia
Incontinentia pigmenti
Wiskott-Aldrich syndrome
Menkes syndrome
Sideroblastic anemia
Aarskog-Scott syndrome
PGK deficiency hemolytic anemia
Pelizaeus-Merzbacher disease
Alport-like hereditary nephritis
Fabry disease
Lowe syndrome
Immunodeficiency, X-linked, with hyper IgM
Lymphoproliferative syndrome
Charcot-Marie-Tooth disease
Choroideremia
Cleft palate, X-linked
Spastic paraplegia, X-linked, uncomplicated
Deafness with stapes fixation
PRPS-related gout
Lesch-Nyhan syndrome
HPRT-related gout
Hemophilia B
Albinism-deafness syndrome
Fragile X syndrome
Hunter syndrome
Hemophilia A
G6PD deficiency; Favism
Drug sensitive anemia
Chronic hemolytic anemia
Manic-depressive illness, X-linked
Colorblindness (several forms)
Dyskeratosis congenita
TKCR syndrome
Adrenoleukodystrophy
Adrenomyeloneuropathy
Emery-Dreifuss muscular dystrophy
Diabetes insipidus, nephrogenic
Myotubular myopathy, X-linked
Otopalatodigital syndrome, type I
X
Chromosome amplified 25%
p
q
Diabetes mellitus, insulin-resistant, with acanthosis nigricans
Leprechaunism
?Rabson-Mendenhall syndrome
C3 deficiency
Leukemia, acute lymphoblastic
Persistent Mullerian duct syndrome
SCID HLA class II negative type
Hypercholesterolemia, familial
Mannosidosis
Central core disease of muscle
Hemolytic anemia due to glucosephosphate isomerase deficiency
Hydrops fetalis
Prolidase deficiency
{Polio, susceptibility to}
Hyperlipoproteinemia, type IB
Hyperlipoproteinemia, type III
Maple syrup urine disease
Myotonic dystrophy
Xeroderma pigmentosum (1 type)
?Male pseudohermaphroditism due to defective LH
?Hypergonadotropic hypogonadism
Leukemia/lymphoma-3, B-cell
Malignant hyperthermia
?Hyperleucine-isoleucinemia
Glutaricaciduria, type IIc
19
Diabetes insipidus, neurohypophyseal
Creutzfeld-Jakob disease
Gerstmann-Straussler disease
[Inosine triphosphatase deficiency]
Arteriohepatic dysplasia
Cerebral amyloid angiopathy
Alagille syndrome
?Growth hormone deficiency due to defect in GHRF
Fanconi anemia-1
MODY, one form
Severe combined immunodeficiency due to adenosine deaminase deficiency
Hemolytic anemia due to ADA excess
Galactosialidosis
Pseudohypoparathyroidism, type Ia
McCune-Albright polyostotic fibrous dysplasia
Pituitary tumor, growth-hormone-secreting
Seizures, benign neonatal
20
Amyotrophic lateral sclerosis, one form
Amyloidosis, cerebroarterial, Dutch type
Alzheimer disease, one form
Leukemia, acute myeloid
Leukocyte adhesion deficiency
Hemolytic anemia due to phosphofructokinase deficiency
Homocystinuria, B6-responsive & B6-unresponsive
Epilepsy, progressive myoclonus
21
Cat eye syndrome*
DiGeorge syndrome*
Alpha-NAGA deficiency
Glutathionuria
Chronic myeloid leukemia (breakpoint)*
Thrombophilia due to heparin cofactor II deficiency
Neuroepithelioma
Ewing sarcoma*
Debrisoquine sensitivity
?Parkinsonism, susceptibility to]
Adenylosuccinase deficiency
Acoustic neuroma *
Meningioma *
Glucose/galactose malabsorption
Transcobalamin II deficiency
Methemoglobinemia (enzymopathic form)
Metachromatic leukodystrophy
22
XY gonadal dysgenesis
Y

Figure 1d

Table 1 The morbid anatomy of the human genome (by chromosome).

Location	Symbol	Status	Title	MIM#	Method	Disorder	Mouse
1pter-p36.13	ENO1, PPH	C	Enolase-1, alpha	172430	S, F, R, REa	Enolase deficiency (1)	4(Eno-1)
1p36.3-p34.1	C1QA	C	Complement component-1, q subcomponent, alpha polypeptide	120550	REa, REb	?C1q deficiency (1)	
1p36.3-p34.1	C1QB	C	Complement component-1, q subcomponent, beta polypeptide	120570	REa, REb	?C1q deficiency (1)	(C1qb)
1p36.3-p36.2	PLOD	P	Procollagen-lysine, 2-oxoglutarate 5-dioxygenase (lysine hydroxylase)	153454	REa, A	Ehlers-Danlos syndrome, type VI, 225400 (1)	
1p36.2-p36.1	NB, NBS	C	Neuroblastoma (neuroblastoma suppressor)	256700	Ch, D	Neuroblastoma (2)	
1p36.2-p34	EL1	C	Elliptocytosis-1 (protein 4.1)	130500	F, REb	Elliptocytosis-1 (3)	4(Elp-1)
1p36.2-p34	EKV	C	Erythrokeratodermia variabilis	133200	F	Erythrokeratodermia variabilis (2)	
1p36.2-p34	RH	C	RHESUS BLOOD GROUP CLUSTER	111700	F, D, Fd, A	Erythroblastosis fetalis (1); ?Rh-null hemolytic anemia (1)	
1p36.1-p34	ALPL, HOPS	C	Alkaline phosphatase, liver/bone/kidney	171760	S, H, Fd, F, A	Hypophosphatasia, infantile 241500 (3); ?Hypophosphatasia, adult 146300 (1)	4(Akp-2)
1p36	BRCD2	P	Breast cancer, ductal	211420	Ch, F, D	Breast cancer, ductal (2)	
1p36	CMM, HCMM, MLM, DNS	I	Malignant melanoma, cutaneous (dysplastic nevus syndrome)	155600	F, Fd	?Malignant melanoma, cutaneous (2)	
1p36-p35	GALE	C	UDP galactose-4-epimerase	230350	S, LD	Galactose epimerase deficiency (1)	
1p34	FUCA1, FUCA	C	Fucosidase, alpha-L- 1, tissue	230000	S, F, R, A, REa	Fucosidosis (1)	4(Fuca)
1p34	UROD	C	Uroporphyrinogen decarboxylase	176100	S, A, REa	Porphyria cutanea tarda (1); Porphyria, hepatoerythropoietic (1)	4(Urod)
1p32	C8A	C	Complement component 8, alpha polypeptide	120950	F, A, Ch	C8 deficiency, type I (2)	
1p32	C8B	C	Complement component 8, beta polypeptide	120960	F, A, Ch, H	C8 deficiency, type II (2)	4(C8b)
1p32	CLN1	P	Ceroid lipofuscinosis, neuronal-1, infantile	256730	Fd, LD	Ceroid lipofuscinosis, neuronal-1, infantile (2)	
1p32	TAL1, TCL5, SCL	C	T-cell leukemia/lymphoma-5 (stem-cell leukemia)	187040	Ch, RE	Leukemia-1, T-cell acute lymphoblastic (3)	4(Scl)
1p31	ACADM, MCAD	P	Acyl-CoA dehydrogenase, medium chain	201450	REa,A	Acyl-CoA dehydrogenase, medium chain, deficiency of (1)	8(Acadm)
1p31	DBT, BCATE2	C	Dihydrolipoamide branched chain transacylase (E2 component of branched chain keto acid dehydrogenase complex)	248610	REa, A	Maple syrup urine disease, type 2 (3)	
1p22.1-q21.1	CD3Z, TCRZ	P	Antigen CD3Z, zeta polypeptide (TiT3 complex)	186780	REa, A	CD3, zeta chain, deficiency (1)	1(T3z)
1p21	AGL	P	Amylo-1,6-glucosidase, 4-alpha-glucanotransferase (glycogen debranching enzyme)	232400	REc, A	Glycogen storage disease-3 (3)	
1p21-p13	AMPD1	P	Adenosine monophosphate deaminase-1	102770	REa, A	Myoadenylate deaminase deficiency (1)	3(Ampd-1)
1p21-p13	AMPD2	L	Adenosine monophosphate deaminase-2	102771	H	[AMP deaminase deficiency, erythrocyte] (1)	3(Ampd-2)
1p21-p13	CSF1, MCSF	C	Colony-stimulating factor-1 (macrophage)	120420	A, REa, H	?Osteopetrosis, 259700 (1)	3(Csfm)
1p13.1	HSD3B2, HSD3B, HSDB3	C	Hydroxy-delta-5-steroid dehydrogenase, 3 beta- and steroid delta-isomerase, type 2	201810	A	Adrenal hyperplasia II (3)	3(Hsd3b)
1p13	TSHB	C	Thyroid-stimulating hormone, beta polypeptide	188540	REa, RE, Fd	Hypothyroidism, nongoitrous (1)	3(Tshb)
1p13-p11	CPT1	C	Carnitine palmitoyltransferase I	255120	REa, A	Myopathy due to CTPase deficiency (1); Hyperammonemia due to CTPase deficiency (1)	
1p11-qter	EPHX, EPOX	P	Epoxide hydroxylase, microsomal (epoxide hydrolase)	132810	REa	?Fetal hydantoin syndrome (1); Diphenylhydantoin toxicity (1)	1(Eph-1)
1cen-q32	PFKM	P	Phosphofructokinase, muscle type	232800	S	Glycogen storage disease VII (1)	
1q	FMO2	P	Flavin-containing monooxygenase 2 (adult liver)	136131	Psh	[Fish-odor syndrome] (1)	
1q2	CAE	C	Cataract, zonular pulverulent	116200	F	Cataract, zonular pulverulent (2)	
1q21	FLG	P	Filaggrin	135940	REa, A	?Ichthyosis vulgaris, 146700 (1)	
1q21	GBA	C	Glucosidase, beta; acid	230800	S, A, D	Gaucher disease (1)	3(Gba)
1q21	PKLR, PK1, PKR	C	Pyruvate kinase, liver and RBC type	266200	REa, A	PK deficiency hemolytic anemia (1)	
1q21	SPTA1	C	Spectrin, alpha, erythrocytic-1	182860	REa, A, Fd	Elliptocytosis-2 (2); Pyropoikilocytosis (1); Spherocytosis, recessive (1)	1(Spna-1)
1q21-q22	FY	C	Duffy blood group	110700	F, Fc, Fd	{Vivax malaria, susceptibility to} (1)	
1q21-q23	APCS, SAP	C	Amyloid P component, serum	104770	REa, A, Fd	{?Amyloidosis, secondary, susceptibility to} (1)	1(Sap)
1q21-q23	NEM1	P	Nemaline myopathy	161800	Fd	Nemaline myopathy (2)	
1q21-q24	GRMP	C	Granulocyte membrane protein (140kD, antigen CD62)	173610	REn, A	Platelet alpha/delta storage pool deficiency (1)	1(Grmp)
1q21.2-q23	CMT1B	C	Charcot-Marie-Tooth disease, slow nerve conduction type Ib	118200	F, Fd, D	Charcot-Marie-Tooth neuropathy, slow nerve conduction type Ib (2)	
1q23	F5	C	Coagulation factor V	227400	REa, A, Fd, REn	Factor V deficiency (1)	1(Cf-5)

Table 1—contd

Location	Symbol	Status	Title	MIM#	Method	Disorder	Mouse
1q23	FCGR3, CD16, IGFR3	C	Fc fragment of IgG, low affinity III, receptor for (CD16) (Immunoglobulin G Fc receptor III)	146740	REb, REn	Lupus erythematosus, systemic, 152700 (1); Neutropenia, immune (2)	
1q23	PBX1	C	Pre-B cell leukemia transcription factor-1	176310	Ch, A	Leukemia, acute pre-B-cell (2)	1(Pbx)
1q23-q25	AT3	C	Antithrombin III	107300	F, D, A, REa, Fd	Antithrombin III deficiency (3)	1(At-3)
1q25	NCF2	C	Neutrophil cytosolic factor-2	233710	REa, A	Chronic granulomatous disease due to deficiency of NCF-2 (1)	1(Ncf-2)
1q31-q32.1	F13B	C	Coagulation factor XIII, B polypeptide	134580	Fd, A, RE	Factor XIIIB deficiency (1)	
1q32	CR1, C3BR	C	Complement component (3b/4b) receptor-1	120620	F, REa, A, RE	CR1 deficiency (1); ?SLE (1)	
1q32	HF, CFH	C	Complement component H	134370	F, REa, RE, H	Factor H deficiency (1)	1(Cfh)
1q32	USH2	C	Usher syndrome-2 (autosomal recessive, mild)	276901	Fd	Usher syndrome, type 2 (2)	
1q32	VWS, LPS, PIT, VDWS	C	Lip pit syndrome (van der Woude syndrome)	119300	Ch, Fd	van der Woude syndrome (2)	
1q41-q42	ADPRT, PPOL, PARP	C	Poly-ADP-ribose polymerase (NAD(+) ADP-ribosyltransferase)	173870	REa, A	?Fanconi anemia (1); ?Xeroderma pigmentosum (1)	
1q42.1	FH	C	Fumarate hydratase	136850	S, R, D	Fumarase deficiency (1)	
Chr.1	HSD11	P	11-beta-hydroxysteroid dehydrogenase	218030	REa	11-beta-hydroxysteroid dehydrogenase deficiency (1)	
2pter-p12	TPO, TPX	C	Thyroid peroxidase	274500	REa, A	Thyroid iodine peroxidase deficiency (1)	12(Tpo)
2p25	POMC	C	Proopiomelanocortin	176830	REa	ACTH deficiency (1)	12(Pomc-1)
2p24	APOB	C	Apolipoprotein B	107730	REa, A	Hypobetalipoproteinemia (1); ?Abetalipoproteinemia (1); Hyperbetalipoproteinemia (1); Apolipoprotein B-100, defective (1)	12(Apob)
2p21	HPC	L	Holoprosencephaly	157170	Ch	?Holoprosencephaly (2)	
2p12	IGKC	C	Constant region of kappa light chain	147200	REa, A	[Kappa light chain deficiency] (1)	
2p	CPS1	P	Carbamoylphosphate synthetase I (mitochondrial CPS)	237300	REa	Carbamoylphosphate synthetase I deficiency (1)	
2q13-q14	PROC	C	Protein C	176860	REa, A	Protein C deficiency (1)	
2q14-q21	LCO, LCA	P	Liver cancer oncogene (oncogene LCA)	165320	REa, REb, A	?Hepatocarcinoma (1)	
2q21	ERCC3, XPB	C	Excision-repair, complementing defective, in Chinese hamster, number 3	133510	S, A	Xeroderma pigmentosum, group B (1)	
2q31	COL3A1	C	Collagen III, alpha-1 polypeptide	120180	REa, A	Ehlers-Danlos syndrome, type IV (3); Aneurysm, familial (1)	1(Col3a-1)
2q31	GAD1	C	Glutamate decarboxylase-1, brain	266100	REa, H, A	?Pyridoxine dependency with seizures (1)	2(Gad-1)
2q31.2	OCA	L	Oculocutaneous albinism, type II (tyrosinase-positive)	203200	Ch	?Oculocutaneous albinism, type II (2)	
2q33-q35	CRYG1	C	Crystallin, gamma polypeptide 1	123660	REa, A	Cataract, Coppock-like (3)	1(Cryg-1)
2q33-qter	CYP27, CTX	P	Sterol 27-hydroxylase	213700	REa	Cerebrotendinous xanthomatosis (2)	1(Cyp27)
2q34	TCL4	P	T-cell leukemia/lymphoma-4	186860	Ch, RE	Leukemia/lymphoma, T-cell (2)	
2q34-q35	ACADL, LCAD	P	Acyl-CoA dehydrogenase, long-chain	201460	A	Acyl-CoA dehydrogenase, long chain, deficiency of (1)	
2q34-q36	FN1	C	Fibronectin-1	135600	S, REa, A	?Ehlers-Danlos syndrome, type X (1)	1(Fn-1)
2q35	DES	P	Desmin	125660	REa, A	?Cardiomyopathy (1)	1(Des)
2q35	PAX3, WS1, HUP2	C	Paired box homeotic gene-3 (Waardenburg syndrome, type I)	193500	Ch, Fd, H, A	Waardenburg syndrome, type I (3)	1(Sp)
2q36-q37	AGXT, AGT, SPAT	P	Alanine:glyoxylate aminotransferase, liver-specific peroxisomal	259900	A, REa	Oxalosis I (1)	
2q36-q37	GCG	C	Glucagon	138030	REa, A	[?Hyperproglucagonemia] (1)	2(Gcg)
2q37	RMSA	C	Rhabdomyosarcoma, alveolar	268220	Ch	Rhabdomyosarcoma, alveolar (2)	
Chr.2	LCT, LAC, LPH	C	Lactase (lactase-phlorizin hydrolase)	223000	REa, Fd	?Lactase deficiency, congenital (1); ?Lactase deficiency, adult, 223100 (1)	
Chr.2	UGT1, GNT1	P	UDP-glucuronosyltransferase-1 family	191740	REa	Crigler-Najjar syndrome, type I, 218800 (1); ? (Gilbert syndrome, 143500) (1)	1(Ugt-1)
3p26-p25	VHL	C	von Hippel-Lindau syndrome	193300	Fd, D	von Hippel-Lindau syndrome (2)	
3p24.3	THRB, THR1, ERBA2	C	Thyroid hormone receptor, beta (oncogene ERBA2)	190160	REa, A, RE, Fd	Thyroid hormone resistance, 274300, 188570 (3)	
3p23-p22	ACAA	P	Peroxisomal 3-oxoacyl-CoA thiolase	261510	REa, A	Pseudo-Zellweger syndrome (1)	
3p23-p21	SCLC1, SCCL	C	Small-cell cancer of lung	182280	Ch, D	Small-cell cancer of lung (2)	
3p21	COL7A1	P	Collagen VII, alpha-1 polypeptide	120120	REa, A	Epidermolysis bullosa dystrophica, dominant, 131750 (3); Epidermolysis bullosa dystrophica, recessive, 226600 (3)	
3p21-p14.2	GLB1	C	Galactosidase, beta-1	230500	S, EM	GM1-gangliosidosis (1); Mucopolysaccharidosis IVB (1)	9(Bgl)

Table 1—contd

Location	Symbol	Status	Title	MIM#	Method	Disorder	Mouse
3p14.2	RCC1, RCC	C	Renal cell carcinoma	144700	Fc, Ch	Renal cell carcinoma (2)	
3p14-qter	HRG	P	Histidine-rich glycoprotein	142640	REa	Thrombophilia due to elevated HRG (2)	
3p11.1-q11.2	PROS1, PSA, PROS	C	Protein S, alpha	176880	REa	Protein S deficiency (1)	
3cen-q22	MER6, RHN	P	Rh-null, regulator type	268150	S	Rh-null disease (1)	
3q	PIT1	P	Pituitary-specific transcription factor Pit-1	173110	Fd	?Panhypopituitarism (1)	16(Pit-1,dw)
3q11-q12	GPX1	C	Glutathione peroxidase-1	138320	S, REa	Hemolytic anemia due to glutathione peroxidase deficiency (1)	
3q13	UMPS, OPRT	C	Orotate phosphoribosyltransferase/OMP decarboxylase (UMP synthase)	258900	S, A	Oroticaciduria (1)	
3q2	BPES	P	Blepharophimosis, epicanthus inversus and ptosis	110100	Ch	Blepharophimosis, epicanthus inversus and ptosis (2)	
3q21	TF	C	Transferrin	190000	S, H, REa, D, A	Atransferrinemia (1)	9(Trf)
3q21-q22	PCCB	C	Propionyl CoA carboxylase, beta polypeptide	232050	REa, A, D	Propionicacidemia, type II or pccB type (1)	
3q21-q23	LTF	C	Lactotransferrin	150210	REa, A	?Lactoferrin-deficient neutrophils, 245480 (1)	9(Ltf)
3q21-q24	CP	C	Ceruloplasmin	117700	F, H, REa, A	[Hypoceruloplasminemia, hereditary] (1)	9(Cp)
3q21-q24	FHH, HHC	P	Familial hypocalciuric hypercalcemia	145980	Fd	Hypercalcemia, hypocalciuric, familial (2)	
3q21-q24	RHO, RP4	C	Rhodopsin	180380	REa, A, Fd	Retinitis pigmentosa, autosomal dominant (3)	6(Rho)
3q25-q26	SI	P	Sucrase-isomaltase	222900	REa, A, Fd	Sucrose intolerance (1)	
3q26-qter	KNG	C	Kininogen	228960	Psh, A	[Kininogen deficiency] (1)	
3q26.1-q26.2	BCHE, CHE1	C	Pseudocholinesterase-1	177400	F, D, A	Postanesthetic apnea (1)	
3q26.3	CDL	L	Cornelia de Lange syndrome	122470	Ch	?Cornelia de Lange syndrome (2)	
Chr.3	TRH	P	Thyrotropin-releasing hormone	275120	REa	Thyrotropin-releasing hormone deficiency (1)	
4p16.3	HD	C	Huntington disease	143100	Fd	Huntington disease (2)	
4p16.3	IDUA, IDA	P	Alpha-L-iduronidase	252800	REa, A, S	Hurler syndrome (1); Mucopolysaccharidosis I (1); Hurler-Scheie syndrome (1); Scheie syndrome (1)	
4p16.1	HOX7	P	Homeo box-7	142983	REa, A, D, Fd	?Wolf-Hirschhorn syndrome (3)	5(Hox-7)
4p15.31	QDPR, DHPR	C	Quinoid dihydropteridine reductase	261630	S, A, REa, D	Phenylketonuria due to dihydropteridine reductase deficiency (1)	5(Qdpr)
4q11-q12	KIT, PBT	C	Hardy-Zuckerman 4 feline sarcoma (v-kit) oncogene	164920	REa, A, H, Ch, H, REn	Piebaldism (3)	5(Kit; W)
4q11-q13	AFP, HPAFP	C	Alpha-fetoprotein	104150	H, A, Fd, F	[AFP deficiency, congenital] (1); [Hereditary persistence of alpha-fetoprotein] (3)	5(Afp)
4q11-q13	ALB	C	Albumin	103600	F, A, REa	Analbuminemia (1); [Dysalbuminemic hyperthyroxinemia] (1); [Dysalbuminemic hyperzincemia] (1)	5(Alb-1)
4q11-q13	JPD, JP	P	Periodontitis, juvenile	170650	F	Periodontitis, juvenile (2)	
4q13-q21	DGI1	C	Dentinogenesis imperfecta-1	125490	F, Fd	Dentinogenesis imperfecta-1 (2)	
4q21	IGJ, IGCJ, JCH	P	J region of immunoglobulin heavy chain	147790	REa, A	?Leukemia, acute lymphocytic, with 4/11 translocation (3)	5(Igj)
4q21-q23	GNPTA	P	N-acetyl-alpha-glucosaminyl-phosphotransferase	252500	F, S, D	Mucolipidosis II (1); Mucolipidosis III (1)	
4q23-q27	AGA	C	Aspartylglucosaminidase	208400	S, F, D	Aspartylglucosaminuria (3)	
4q23-q27	RGS	P	Rieger syndrome	180500	Ch, Fd	Rieger syndrome (2)	
4q25	IF	C	Complement component I (C3b inactivator)	217030	REa, Fd, A, RE	C3b inactivator deficiency (1)	
4q26-q27	IL2, TCGF	C	T-cell growth factor (interleukin-2)	147680	REa, A, F	Severe combined immunodeficiency due to IL2 deficiency (1)	3(Il-2)
4q28	FGA	C	Fibrinogen, alpha chain	134820	RE, REa, H, D, LD, A	Dysfibrinogenemia, alpha types (1)	
4q28	FGB	C	Fibrinogen, beta chain	134830	RE, REa, D, LD, A	Dysfibrinogenemia, beta types (1)	
4q28	FGG	C	Fibrinogen, gamma chain	134850	F, REa, H, RE, D, LD, A	Dysfibrinogenemia, gamma types (1); Hypofibrinogenemia, gamma types (1)	3(Fgg)
4q28-q31	ASMD	P	Anterior segment mesenchymal dysgenesis	107250	F	Anterior segment mesenchymal dysgenesis (2)	
4q28-q31	TYS	C	Sclerotylosis	181600	F	Sclerotylosis (2)	
4q31.1	MLR, MCR, MR	C	Mineralocorticoid receptor	264350	REa, M, A	Pseudohypoaldosteronism (1)	
4q32.1	HVBS6, HCC2	P	Hepatitis B virus integration site-6 (hepatocellular carcinoma-2)	142380	REa, A, D	Hepatocellular carcinoma (3)	
4q35	F11	C	Coagulation factor XI	264900	A, H, Fd	Factor XI deficiency (1)	8(cf-11)
4q35	FMD, FSHD	C	Facioscapulohumeral muscular dystrophy	158900	Fd	Facioscapulohumeral muscular dystrophy (2)	
4q35	KLK3	P	Kallikrein, plasma	229000	A	Fletcher factor deficiency (1)	8(Kal-3)
5p13	C6	C	Complement component-6	217050	A, H, RE, Fd, LD	C6 deficiency (1); Combined C6/C7 deficiency (1)	15(C6)
5p13	C7	C	Complement component-7	217070	A, H, RE, Fd, LD	C7 deficiency (1)	15(C7)

Table 1—contd

Location	Symbol	Status	Title	MIM#	Method	Disorder	Mouse
5p13	C9	C	Complement component-9	120940	REa, A, Fd, LD	C9 deficiency (1)	
5p13-p12	GHR, GHBP	C	Growth hormone receptor	262500	REa, A	Laron dwarfism (1)	15(Ghr)
5p13-p12	BBBG	L	Hypospadias-dysphagia syndrome (Opitz BBBG syndrome)	145410	Ch	?Hypospadias-dysphagia syndrome (2)	
5q11-q13	ARSB	C	Arylsulfatase B	253200	S	Maroteaux-Lamy syndrome, several forms (1)	13(As-1)
5q11.2-q13.2	DHFR	C	Dihydrofolate reductase	126060	S, REa, H, D	?Anemia, megaloblastic, due to DHFR deficiency (1)	13(Dhfr)
5q11.2-q13.3	SCZD1	L	Schizophrenia-1	181510	Ch, Fd	?Schizophrenia (2)	
5q12-q32	MAR	P	Macrocytic anemia, refractory	153550	Ch	Macrocytic anemia of 5q- syndrome, refractory (2)	
5q12.2-q13.3	SMA	C	Spinal muscular atrophy	253300	Fd	Werdnig-Hoffmann disease (2); Spinal muscular atrophy II (2); Spinal muscular atrophy III (2)	
5q13	HEXB	C	Hexosaminidase B (beta polypeptide)	268800	S, Ch, D	Sandhoff disease (1)	13(Hex-2)
5q21	MCC	C	Mutated in colorectal cancers	159350	REn, D	Colorectal cancer (1)	18(Mcc)
5q21-q22	APC, GS, FPC	C	Adenomatous polyposis of the colon (Gardner syndrome; familial polyposis coli)	175100	D, Fd, REn	Gardner syndrome (3); Polyposis coli, familial (3); Colorectal cancer (3)	18(Min)
5q22.3-q31.3	LGMD1	P	Limb-girdle muscular dystrophy, autosomal dominant	159000	Fd	Muscular dystrophy, limb-girdle, autosomal dominant (2)	
5q23	DTS	C	Diphtheria toxin sensitivity	126150	S, M	{Diphtheria, susceptibility to} (1)	
5q31	GRL	C	Glucocorticoid receptor, lymphocyte	138040	S, REa, Fd, H, A, D, REn	Cortisol resistance (1)	18(Grl-1)
5q31-q33	LFHL1	P	Progressive low-frequency deafness	124900	Fd	Deafness, low-tone (2)	
5q31-q34	DTD, DD	P	Diastrophic dysplasia	222600	Fd	Diastrophic dysplasia (2)	
5q31.3-q33.3	TCOF1, TCS, MFD1	C	Treacher Collins mandibulofacial dysostosis	154500	Ch, Fd	Treacher Collins mandibulofacial dysostosis (2)	
5q33-q35	STHE	P	Startle disease/hyperekplexia (stiff man syndrome)	149400	Fd	Startle disease (2)	
5q33-qter	F12, HAF	C	Coagulation factor XII (Hageman factor)	234000	REa, A	Factor XII deficiency (1)	
Chr.5	FBN2, CCA	P	Fibrillin-2	121050	REa, Fd	Contractural arachnodactyly, congenital (3)	
Chr.5	GM2A	C	GM2-activator protein	272750	S, REa	GM2-gangliosidosis, AB variant (1)	
6pter-p23	OFC, CL	P	Orofacial cleft (cleft lip with or without cleft palate; isolated cleft palate)	119530	F	Orofacial cleft (2)	
6p25-p24	F13A1, F13A	C	Coagulation factor XIII, A polypeptide	134570	F, Fd, A, D	Factor XIIIA deficiency (3)	
6p22-p21	BCKDHB, E1B	C	Branched-chain keto acid dehydrogenase E1, beta polypeptide	248611	REa, A	Maple syrup urine disease, type 3 (1)	
6p21.3	ASD2	P	Atrial septal defect, secundum type	108800	F	Atrial septal defect, secundum type (2)	
6p21.3	C2	C	Complement component-2	217000	F, LD, RE	C2 deficiency (3)	17(C2)
6p21.3	C4A, C4S	C	Complement component-4A	120810	F, H, RE, Fd	C4 deficiency (3)	17(C4)
6p21.3	C4B, C4F	C	Complement component-4B	120820	F, H, RE, Fd	C4 deficiency (3)	17(C4)
6p21.3	CYP21, CA21H, CAH1	C	Cytochrome P450, subfamily XXI; steroid 21-hydroxylase, congenital adrenal hyperplasia	201910	F, RE	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency (3)	17(Cyp21)
6p21.3	EJM1, JME	C	Epilepsy, juvenile myoclonic	254770	F, Fd	Epilepsy, juvenile myoclonic (2)	
6p21.3	GLUR	P	Renal glucosuria	233100	F	[Renal glucosuria] (2)	
6p21.3	HFE	C	Hemochromatosis	235200	LD, F	Hemochromatosis (2)	
6p21.3	IDDM	L	Insulin dependent diabetes mellitus	222100	F, LD	?Diabetes mellitus, insulin-dependent (2)	
6p21.3	NDF	L	Neutrophil differentiation factor	202700	LD	?Kostmann agranulocytosis (2)	
6p21.3	NEU, NEU1	I	Neuraminidase-1; sialidosis	256550	H, F	?Sialidosis (2)	17(Neu-1)
6p21.3	PDB	L	Paget disease of bone	167250	F	?Paget disease of bone (2)	
6p21.3	RWS	L	Ragweed sensitivity	179450	F	?Ragweed sensitivity (2)	
6p21.3-p21.2	LAP	L	Laryngeal adductor paralysis	150270	F	?Laryngeal adductor paralysis (2)	
6p21.3-p21.2	SCA1	C	Spinocerebellar ataxia-1	164400	F, Fd	Spinocerebellar ataxia-1 (2)	
6p21.1-cen	RDS	C	Retinal degeneration, slow (peripherin)	179605	REa, A	Retinitis pigmentosa, peripherin-related (3)	17(rds)
6p21	MUT, MCM	C	Methylmalonyl CoA mutase	251000	REa, A, F, D	Methylmalonicaciduria, mutase deficiency type (1)	17(Mut)
6p	ICS	L	Immotile cilia syndrome	242650	F	?immotile cilia syndrome (2)	
6q13-q15	OAR	L	Ocular albinism, autosomal recessive	203310	Ch	?Ocular albinism autosomal recessive (2)	
6q13-q21	MCDR1, NCMD	P	Macular dystrophy, retinal, 1 (North Carolina type)	136550	Fd	Macular dystrophy, North Carolina type (2)	
6q23	ARG1	P	Arginase, liver	207800	REa	Argininemia (1)	
6q24-q27	ESR, ER	C	Estrogen receptor	133430	REa, A	Breast cancer (1)	
6q25-q26	RCD1	L	Retinal cone dystrophy-1	180020	Ch	?Retinal cone dystrophy-1 (2)	
6q26-q27	PLG	C	Plasminogen	173350	REa, A, LD, F	Plasminogen Tochigi disease (1); Dysplasminogenemic thrombophilia (1); Plasminogen deficiency, types I and II (1)	17(Plg)
6q27	LPA	C	Apolipoprotein Lp(a)	152200	REa, A, F, Fd	{Coronary artery disease, susceptibility to} (1)	
7p21.3-p21.2	CRS, CSO	C	Craniosynostosis	123100	Ch	Craniosynostosis (2)	
7p15-p13	GCK	P	Glucokinase	138079	Psh, Fd	MODY, type II, 125851 (3)	
7p13	GCPS	C	Greig craniopolysyndactyly syndrome	175700	Ch, Fd, REn, D	Greig craniopolysyndactyly syndrome (3)	13(Xt)

Table 1—contd

Location	Symbol	Status	Title	MIM#	Method	Disorder	Mouse
7p13-p12.3	PGAM2, PGAMM	C	Phosphoglycerate mutase, muscle form	261670	REa, A	Myopathy due to phosphoglycerate mutase deficiency (1)	
7p	GHS	L	Goldenhar syndrome	141400	Ch	?Goldenhar syndrome (2)	
7cen-q11.2	ASL	C	Argininosuccinate lyase	207900	S, REa, A	Argininosuccinicaciduria (1)	5(Asl)
7q11.2-q21.3	EEC	L	Ectrodactyly, ectodermal dysplasia, cleft lip/palate	129900	Ch	?EEC syndrome (2)	
7q11.23	NCF1	P	Neutrophil cytosolic factor-1	233700	REa, A	Chronic granulomatous disease due to deficiency of NCF-1 (1)	
7q11.23	ZWS1, ZWS, ZS	C	Zellweger syndrome-1	214100	Ch	Zellweger syndrome-1 (2)	
7q21	EPO	C	Erythropoietin	133170	REa, A, REb, Fd	?Erythremia (1)	5(Epo)
7q21.11	GUSB	C	Beta-glucuronidase	253220	S, D, EM	Mucopolysaccharidosis VII (1)	5(Gus)
7q21.2-q21.3	SHFD1	C	Split-hand/foot deformity, type 1	183600	Ch	Split-hand/foot deformity, type 1 (2)	
7q21.3-q22	PLANH1, PAI1	C	Plasminogen activator inhibitor, type I	173360	REa, REb, Fd, A, D	Thrombophilia due to excessive plasminogen activator inhibitor (1); Hemorrhagic diathesis due to PAI1 deficiency (1)	
7q21.3-q22.1	COL1A2	C	Collagen I, alpha-2 polypeptide	120160	S, REa, D, A	Osteogenesis imperfecta, 2 or more clinical forms (3); Ehlers-Danlos syndrome, type VIIA2 (3)	6(Cola-2)
7q22-qter	BCP, CBT	P	Blue cone pigment	190900	REa	Colorblindness, tritan (2)	
7q31	OB	L	Obesity	164160	H	?Obesity (2)	6(ob)
7q31-q32	DLD, LAD, PHE3	C	Dihydrolipoamide dehydrogenase (pyruvate dehydrogenase component E3)	246900	REa	Lipoamide dehydrogenase deficiency (1)	
7q31-q34	BPGM	P	2,3-bisphosphoglycerate mutase	222800	A	Hemolytic anemia due to bisphosphoglycerate mutase deficiency (1)	
7q31.1-q31.3	LAMB1	C	Laminin B1	150240	REa, A, Ch	?Cutis laxa, marfanoid neonatal type (1)	1(Lamb-1)
7q31.2	CFTR, CF	C	Cystic fibrosis transmembrane conductance regulator	219700	F, Fd	Cystic fibrosis (3); congenital absence of vas deferens (1)	6(Cftr)
7q32-qter	TRY1, TRP1	P	Trypsin-1	276000	REa	Trypsinogen deficiency (1)	6(Try-1)
7q34-qter	SLO	L	Smith-Lemli-Opitz syndrome	270400	Ch	?Smith-Lemli-Opitz syndrome (2)	
7q36	HLP3	P	Holoprosencephaly, type 3	142945	Ch	Holoprosencephaly, type 3 (2)	
7q36	HPFH2	L	Hereditary persistence of fetal hemoglobin, heterocellular, Indian type	142335	Fd	?Hereditary persistence of fetal hemoglobin, heterocellular, Indian type (2)	
Chr.7	HADH	P	Hydroxyacyl-CoA dehydrogenase	143450	S	3-hydroxyacyl-CoA dehydrogenase deficiency (1)	
8p22	LPL, LIPD	P	Lipoprotein lipase (lipase D)	238600	REa, A	Hyperlipoproteinemia I (1)	8(Lpl)
8p21.1	GSR	C	Glutathione reductase	138300	S, D	Hemolytic anemia due to glutathione reductase deficiency (1)	8(Gr-1)
8p21-p11.2	LHRH, GNRH	P	Luteinizing hormone releasing hormone (gonadotropin releasing hormone)	152760	REa, A	?Hypogonadotropic hypogonadism due to GNRH deficiency, 227200 (1)	14(Gnrh)
8p12	PLAT, TPA	C	Plasminogen activator, tissue type	173370	REa, A, REb	Plasminogen activator deficiency (1)	8(Plat)
8p12-p11	WRN	P	Werner syndrome	277700	Fd	Werner syndrome (2)	
8p11.2	ANK1, SPH2	C	Ankyrin-1, erythrocytic	182900	F, Ch, D, REa, A, Fd, REb	Spherocytosis-2 (3)	8(nb)
8p11-q21	RP1	P	Retinitis pigmentosa-1	180100	Fd	Retinitis pigmentosa-1 (2)	
8q12	SGPA, PSA	P	Salivary gland pleomorphic adenoma	181030	Ch	Salivary gland pleomorphic adenoma (2)	
8q13.3	BOR, BOS	C	Branchiootic syndrome	113650	Ch, Fd	Branchiootic syndrome (2)	
8q21	CYP11B1, P450C11	C	Cytochrome P450, subfamily XIB, polypeptide-1; 11-beta-hydroxylase; corticosteroid methyl-oxidase II (CMO II)	202010	REa, A, Ch	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency (1); CMO II deficiency (1)	
8q21	CYP11B2	C	Cytochrome P450, subfamily XIB, polypeptide-2	124080	REa	CMO II deficiency (1)	
8q21.4	CMD1	L	Campomelic dysplasia with sex reversal	211970	Ch	?Campomelic dysplasia with sex reversal (2)	
8q22	CA1	C	Carbonic anhydrase I	114800	REa, H, A	[Carbonic anhydrase I deficiency] (1)	3(Car-1)
8q22	CA2	C	Carbonic anhydrase II	259730	REa, H	Renal tubular acidosis-osteopetrosis syndrome (1)	3(Car-2)
8q22	CLCD	L	Cleidocranial dysplasia	119600	Ch	?Cleidocranial dysplasia (2)	
8q23-q24.1	EXT	L	Multiple exostoses	133700	Ch	?Multiple exostoses (2)	
8q24	EBS1	C	Epidermolysis bullosa simplex-1 (Ogna)	131950	F	Epidermolysis bullosa, Ogna type (2)	
8q24	PDS	L	Pendred syndrome	274600	Ch	?Pendred syndrome (2)	
8q24	VMD1	C	Macular dystrophy, atypical vitelliform	153840	F	Macular dystrophy, atypical vitelliform (2)	
8q24.11-q24.13	LGCR, LGS, TRPS2	C	Langer-Giedion syndrome	150230	Ch	Langer-Giedion syndrome (2)	
8q24.12	TRPS1	P	Trichorhinophalangeal syndrome, type I	190350	Ch	Trichorhinophalangeal syndrome, type I (2)	
8q24.12-q24.13	MYC	C	Oncogene MYC, avian myelocytomatosis virus	190080	REa, A	Burkitt lymphoma (3)	15(Myc)
8q24.2-q24.3	TG	C	Thyroglobulin	188450	A, REa, REb	Hypothyroidism, hereditary congenital (1); Goiter, adolescent multinodular (1)	15(Tgn; cog)

Table 1—contd

Location	Symbol	Status	Title	MIM#	Method	Disorder	Mouse
Chr.8	RTS	L	Rothmund-Thomson syndrome	268400	Ch	?Rothmund-Thomson syndrome (2)	
9p24	OVC	P	Oncogene OVC (ovarian adenocarcinoma oncogene)	164759	Ch	Ovarian carcinoma (2)	
9p23	TYRP, CAS2, CATB	C	Tyrosinase-related protein (catalase B; human homolog of 'brown' locus)	115501	Psh, REa, A	?Melanoma (1)	4(b;trp-1)
9p22	NKH1	L	Hyperglycinemia, isolated nonketotic, type I	238300	Ch	?Hyperglycinemia, isolated nonketotic, type I (2)	
9p22-p21	LALL	P	Lymphomatous acute lymphoblastic leukemia	247640	Ch	Leukemia, acute lymphoblastic (2)	
9p21	IFNA, IFL,IFA	C	Interferon, alpha (leukocyte)	147660	REa, A, RE	Interferon, alpha, deficiency (1)	4(Ifa)
9p13	GALT	C	Galactose-1-phosphate uridyltransferase	230400	S, D, F	Galactosemia (1)	4(Galt)
9q13-q21.1	FRDA, FAT	C	Friedreich ataxia	229300	Fd	Friedreich ataxia (2)	
9q22	ALDOB	C	Aldolase B, fructose-bisphosphatase	229600	REb, REa, A, D	Fructose intolerance (1)	
9q31	ESS1, MSSE	P	Epithelioma, self-healing, squamous 1, Ferguson-Smith type	132800	Fd	Epithelioma, self-healing, squamous 1, Ferguson-Smith type (2)	
9q31	BCNS, NBCCS	C	Basal cell nevus syndrome (Gorlin syndrome)	109400	Fd, D	Basal cell nevus syndrome (2)	
9q31	TAL2	P	T-cell acute lymphoblastic leukemia-2	186855	REa, A, RE, Ch	Leukemia-2, T-cell acute lymphoblastic (3)	
9q32-q33	AMBP, ITIL, ITI, HCP	C	Alpha-1-microglobulin/bikunin; inter-alpha-trypsin inhibitor, light chain; protein HC	176870	REa, A, H	?Familial Mediterranean fever, 249100 (1)	4(Intin-4)
9q32-q34	DYT1	C	Torsion dystonia, autosomal dominant	128100	Fd	Torsion dystonia (2)	
9q33-q34	TSC1, TSC, TS	C	Tuberous sclerosis-1	191100	F, Fd	Tuberous sclerosis-1 (2)	
9q33-qter	ITO	I	Hypomelanosis of Ito	146150	X/A	?Hypomelanosis of Ito (2)	
9q34	ALAD	C	Delta-aminolevulinate dehydratase	125270	F, S, A, REa	Porphyria, acute hepatic (1); Lead poisoning, susceptibility to (1)	4(Lv)
9q34	ASS	C	Argininosuccinate synthetase	215700	S, D, REa, Fd	Citrullinemia (1)	2(Ass-1)
9q34	DBH	C	Dopamine-beta-hydroxylase	223360	F, A	Autonomic failure due to DBH deficiency (1)	2(Dbh)
9q34	GSN	P	Gelsolin	137350	A, REa, RE	Amyloidosis, Finnish type, 105120 (1)	2(Gsn)
9q34	NPS1	C	Nail-patella syndrome	161200	F, Fd	Nail-patella syndrome (2)	
9q34.1	ABL	C	Oncogene ABL (Abelson strain, murine leukemia virus)	189980	REa, Ch, A	Leukemia, chronic myeloid (3)	2(Abl)
9q34.1	AK1	C	Adenylate kinase-1, soluble	103000	F, S, D, Fc	Hemolytic anemia due to adenylate kinase deficiency (1)	2(Ak-1)
9q34.1	C5	C	Complement component-5	120900	REa, A	C5 deficiency (1)	2(Hc)
9q34.1	XPA	C	Xeroderma pigmentosum, complementation group A	278700	S, A	Xeroderma pigmentosum, type A (1)	4(Xpa)
9q34.3	TAN1	P	Translocation-associated 'Notch' homolog	190198	Ch	Leukemia, T-cell acute lymphoblastic (2)	
Chr.9	CPO, CPRO	P	Coproporphyrinogen oxidase	121300	S	Coproporphyria (1); Harderoporphyrinuria (1)	
10p12-q23.2	GBM	C	Glioblastoma multiforme	137800	D	Glioblastoma multiforme (2)	
10q11.2-q21	MBL	C	Mannose-binding lectin	154545	REa, A, Fd	Chronic infections (1)	
10q11-q12	D10S170, TST1, PTC, TPC	C	DNA segment, single copy, probe pH4 (transforming sequence, thyroid-1, from papillary thyroid carcinoma)	188550	REa, A	Thyroid papillary carcinoma (1)	
10q21-q22	PSAP, SAP1, SAP2	C	Prosaposin (sphingolipid activator protein-1; sphingolipid activator protein-2)	176801	S, REa, A, D	Metachromatic leukodystrophy due to deficiency of SAP-1 (1); Gaucher disease, variant form (1)	
10q21.1	CS1	L	Cockayne syndrome, late-onset form	216400	Ch	?Cockayne syndrome (2)	
10q21.1	MEN2A, MEN2	C	Multiple endocrine neoplasia, type II (or IIA); medullary thyroid carcinoma	171400	Fd	Multiple endocrine neoplasia II (2); Medullary thyroid carcinoma (2)	
10q21.1	MEN2B, MEN3	C	Multiple endocrine neoplasia, type IIB (or III)	162300	Fd	Multiple endocrine neoplasia III(2)	
10q22	HK1	C	Hexokinase-1	142600	S, D, A, REa	Hemolytic anemia due to hexokinase deficiency (1)	10(Hk-1)
10q23-q24	RBP4	C	Retinol-binding protein-4, interstitial	180250	REa, A	?Retinol binding protein, deficiency of (1)	19(Rbp-4)
10q24	HOX11, TCL3	P	Homeo box-11 (T-cell leukemia-3 associated breakpoint)	186770	Ch	Leukemia, T-cell acute lymphocytic (2)	
10q24-q25	LIPA	C	Lysosomal acid lipase-A	278000	S, H	Wolman disease (1); Cholesteryl ester storage disease (1)	19(Lip-1)
10q25.2-q26.3	UROS	P	Uroporphyrinogen III synthase	263700	REa, Psh	Porphyria, congenital erythropoietic (1)	
10q26	OAT	C	Ornithine aminotransferase	258870	S, REa, A, Fd	Gyrate atrophy of choroid and retina with ornithinemia, B6 responsive or unresponsive (1)	7(Oat)
10q26.1	PNLIP	P	Lipase, pancreatic	246600	REa, A	Pancreatic lipase deficiency (1)	
Chr.10	CYP17, P450C17	P	Cytochrome P450, subfamily XVII; steroid 17-alpha-hydroxylase	202110	REa, H	Adrenal hyperplasia V (1)	19(Cyp17)
11pter-p15.4	BWS, BWCR, WBS	C	Beckwith-Wiedemann syndrome	130650	Ch, Fd	Beckwith-Wiedemann syndrome (2)	

Table 1—contd

Location	Symbol	Status	Title	MIM#	Method	Disorder	Mouse
11pter-p12	SAA	C	Serum amyloid A	104750	REa, H	?Susceptibility to amyloid in FMF, 249100 (1)	7(Saa-1)
11p15.5	ADCR, ADCC	P	Adrenocortical carcinoma region	202300	D	Adrenocortical carcinoma (2)	
11p15.5	HBB	C	Hemoglobin beta	141900	LD, AAS, F, Fd	Sickle cell anemia (1); Thalassemias, beta- (1); Methemoglobinemias, beta- (1); Erythremias, beta- (1); Heinz body anemias, beta- (1); HPFH, deletion type (1)	7(Hbb)
11p15.5	HBGR	C	Hb gamma regulator	142270	RE	?Hereditary persistence of fetal hemoglobin (3)	
11p15.5	HBG1	C	Hemoglobin gamma 136 alanine	142200	RE	HPFH, nondeletion type A (1)	
11p15.5	HBG2	C	Hemoglobin gamma 136 glycine	142250	RE	HPFH, nondeletion type G (1)	
11p15.5	INS	C	Insulin	176730	HS, A, REb, Fd, D	Diabetes mellitus, rare form (1); MODY, one form, 125850 (3); Hyperproinsulinemia, familial (1)	6(Ins-1); 7(Ins-2)
11p15.5	LQT	P	Long QT syndrome (Romano-Ward syndrome)	192500	Fd	Long QT syndrome (2)	
11p15.5	MAFD1, MD1	L	Manic-depressive illness (major affective disorder 1)	125480	Fd	?Manic-depressive illness (2)	
11p15.5	MTACR1, WT2	P	Multiple tumor associated chromosome region-1	194071	D	Wilms tumor, type 2 (2)	
11p15.5	RMS, RMSCR, RMS1, RMSE	P	Rhabdomyosarcoma, embryonal	268210	D	Rhabdomyosarcoma (2)	
11p15.4	LDHA	C	Lactate dehydrogenase A	150000	S, D, REb, C, A	Exertional myoglobinuria due to deficiency of LDH-A (1)	7(Ldh-1)
11p15.4-15.1	SMPD1, NPD	P	Sphingomyelinase (Niemann-Pick disease)	257200	REa, A	Niemann-Pick disease (1)	
11p15.3-p15.1	PTH	C	Parathyroid hormone	168450	REa, REb, A, Fd	Hypoparathyroidism, familial (1)	7(Pth)
11p15	FCP, HPFH, HHPF	L	F-cell production	142470	F	Heterocellular hereditary persistence of fetal hemoglobin (2)	
11p15	RBTN1, RHOM1	C	Rhombotin-1	186921	Ch, D	Leukemia, T-cell acute lymphoblastic (2)	
11p14-p13	CD59	P	Antigen CD59 (p18-20)	107271	REa	Paroxysmal nocturnal hemoglobinuria (1)	15(Ly-6)
11p14-p13	HVBS1, HBVS1	C	Hepatitis B virus integration site-1	114550	REa, A, Ch	Liver cell carcinoma (1)	
11p13	CAT	C	Catalase	115500	S, D, Fd	Acatalasemia (1)	2(Cas-1)
11p13	FSHB	C	Follicle-stimulating hormone, beta polypeptide	136530	D, REa	?Male infertility, familial (1)	2(Fshb)
11p13	GUD	P	Genitourinary dysplasia	137357	Ch	Genitourinary dysplasia (2)	
11p13	PAX6, AN2	C	Paired box homeotic gene-6	106210	Ch, Fd	Aniridia-2 (3)	2(Sey)
11p13	TCL2	P	T-cell leukemia/lymphoma-2	151390	Ch, RE, A, REa	Leukemia, acute T-cell (2)	
11p13	WT1, WAGR, WTCR1	C	Wilms tumor 1 (Wilms tumor, aniridia, gonadoblastoma, retardation complex)	194070	Ch	Wilms tumor (2); Aniridia of WAGR syndrome (2); Gonadoblastoma (2); Mental retardation of WAGR (2)	2(Wt-1)
11p12-p11	ACP2	C	Acid phosphatase-2	171650	S, REa	?Lysosomal acid phosphatase deficiency (1)	2(Acp-2)
11p11-q12	F2	C	Prothrombin (clotting factor II)	176930	REa, A	Hypoprothrombinemia (1); Dysprothrombinemia (1)	2(Cf-2)
11q	JBS	L	Jacobsen syndrome	147791	Ch	?Jacobsen syndrome (2)	
11q	PC	P	Pyruvate carboxylase	266150	REa, H	Pyruvate carboxylase deficiency (1)	19(Pc)
11q	PORC	P	Porphyria, Chester type	176010	Fd	Porphyria, Chester type (2)	
11q11-q13.1	C1NH, C1I, HANE	C	Complement component-1 inhibitor	106100	REa, A	Angioedema, hereditary (1)	
11q12-q13	APY, IGEL	C	Atopy (allergic asthma and rhinitis; immunoglobulin E level)	147050	Fd	Atopy (2)	
11q13	CCND1, PRAD1, D11S287E	C	Cyclin D1	168461	REn, R, REa, A	Parathyroid adenomatosis 1 (2); Centrocytic lymphoma (2)	
11q13	MEN1	C	Multiple endocrine neoplasia, type I	131100	Fd, D	Multiple endocrine neoplasia I (1)	
11q13	PYGM, MGP	C	Phosphorylase, glycogen, muscle	232600	REb, Fd	McArdle disease (1)	19(Pygm)
11q13	ST3, TSHL	C	Suppression of tumorigenicity-3 (tumor-suppressor gene, HELA cell type)	191181	S, D	Cervical carcinoma (2)	
11q13	VMD2	P	Vitelliform macular dystrophy (Best disease)	153700	Fd	Macular dystrophy, vitelliform type (2)	
11q13-q23	EVR, FEVR	P	Exudative vitreoretinopathy, familial (Criswick-Schepens syndrome)	133780	Fd	Vitreoretinopathy, exudative, familial (2)	
11q13.3	BCL1	C	B-cell CLL/lymphoma-1	151400	RE, Ch	Leukemia/lymphoma, B-cell, 1 (2)	
11q14-q21	TYR, ATN	C	Tyrosinase (albinism, tyrosinase negative)	203100	REa, A, H, F	Albinism (3)	7(Tyr)
11q22-q23	ATA, AT1	C	Ataxia-telangiectasia (complementation groups A, C, D)	208900	Fd, C, M	Ataxia-telangiectasia (2)	
11q22-qter	ANC	L	Anal canal carcinoma	105580	Ch	?Anal canal carcinoma (2)	
11q22.3-q23.1	ACAT	P	Acetyl-Coenzyme A acetyltransferase (acetoacetyl Coenzyme A thiolase)	203750	A	3-ketothiolase deficiency (1)	

Table 1—contd

Location	Symbol	Status	Title	MIM#	Method	Disorder	Mouse
11q23	APOA1	C	Apolipoprotein A-I	107680	REa, RE, Fd, F, D	ApoA-I and apoC-III deficiency, combined (1); Hypertriglyceridemia, one form (1); Hypoalphalipoproteinemia (1); Amyloidosis, Iowa type, 107680.0010 (2)	9(Apoa-1)
11q23	MLL	P	Myeloid/lymphoid, or mixed-lineage leukemia	159555	Ch, RE	Leukemia, myeloid/lymphoid or mixed-lineage (2)	
11q23	TSC2	L	Tuberous sclerosis-2	191090	Ch, Fd	?Tuberous sclerosis-2 (2)	
11q23-qter	PGL, CBT1	P	Paraganglioma (carotid body tumors)	168000	Fd	Paraganglioma (2)	
11q24.1-q24.2	PBGD, UPS	C	Porphobilinogen deaminase (uroporphyrinogen I synthase)	176000	S, D	Porphyria, acute intermittent (1)	9(Ups)
Chr.11	GLAU1	L	Congenital glaucoma-1	231300	Ch	Glaucoma, congenital (2)	
Chr.11	GIF	P	Gastric intrinsic factor	261000	REa	Anemia, pernicious, congenital, due to deficiency of intrinsic factor (1)	
12pter-p12	F8VWF, VWF	C	Coagulation factor VIII VWF (von Willebrand factor)	193400	A, REa, REb, Fd	von Willebrand disease (1)	
12pter-q12	BCT1	C	Branched-chain aminotransferase-1	113520	S	?Hyperleucinemia-isoleucinemia or hypervalinemia (1)	
12p13.3-p12.3	A2M	C	Alpha-2-macroglobulin	103950	REa, A	Emphysema due to alpha-2-macroglobulin deficiency (1)	
12p13	C1R	C	Complement component-1, r subcomponent	216950	REa, Fd, RE, A	C1r/C1s deficiency, combined (1)	
12p13	C1S	C	Complement component-1, s subcomponent	120580	REa, Fd, RE, A	C1r/C1s deficiency, combined (1)	
12p13	MPE, EMP	L	Eosinophils, malignant proliferation of	131440	Ch	?Eosinophilic myeloproliferative disorder (2)	
12p13	TPI1,TPI	C	Triosephosphate isomerase	190450	S, D, R, REa	Hemolytic anemia due to triosephosphate isomerase deficiency (1)	6(Tpi-1)
12p12.1	KRAS2, RASK2	C	Kirsten rat sarcoma-2 viral (v-Ki-ras2) oncogene homolog	190070	REa, A, Fd	Colorectal adenoma (1); Colorectal cancer (1)	6(Kras-2)
12p12.1-p11.2	PTHLH	P	Parathyroid hormone-like hormone	168470	REa, A	?Humoral hypercalcemia of malignancy (1)	6(Pthlh)
12q11-q13	KRT1, KRTA	C	Keratin-1, alpha	139350	H, REa, A	Epidermolysis bullosa simplex, generalized, 131900 (1); Epidermolytic hyperkeratosis, 113800 (2); ?Epidermolysis bullosa simplex, localized, 131800 (1)	15(Krt-2)
12q11-q13	KRT5	P	Keratin-5	148040	A	Epidermolysis bullosa simplex, Dowling-Meara type, 131760 (3); ?Epidermolysis bullosa, Weber Cockayne type, 131800 (2)	
12q12-q14	VDR	P	Vitamin D (1,25-dihydroxyvitamin D3) receptor	277440	REa, A	Rickets, vitamin D-resistant (1)	
12q13-q14	BABL, LIPO	C	Lipoma (breakpoint in benign lipoma); myxoid liposarcoma	151900	Ch	Lipoma (2); Myxoid liposarcoma (2); ?Multiple lipomatosis (2)	
12q13.11-q13.2	COL2A1	C	Collagen II, alpha-1 polypeptide	120140	REa, A	Stickler syndrome (3); Spondyloepiphyseal dysplasia congenita (3); ?Kniest dysplasia (1); Langer-Saldino achondrogenesis-hypochondrogenesis (1); Osteoarthrosis, precocious (3)	
12q14	GNS, G6S	P	N-acetylglucosamine-6-sulfatase	252940	A, REa	Sanfilippo syndrome D (1)	10(Gli)
12q14	PDDR, VDD1	C	Pseudo-vitamin D dependency rickets 1	264700	Fd	Pseudo-vitamin D dependency rickets 1 (2)	
12q22-q23	HAL, HSTD	C	Histidine ammonia-lyase (histidase)	235800	REa, A	[Histidinemia] (1)	10(Hstd)
12q22-qter	ACADS	P	Acyl-CoA dehydrogenase, short chain	201470	REa	Acyl-CoA dehydrogenase, short chain, deficiency of (1)	5(Bcd-1)
12q23.3	TSC3	L	Tuberous sclerosis-3	191091	Fd, Ch	?Tuberous sclerosis-3 (2)	
12q24.1	IFNG, IFI, IFG	C	Interferon, gamma or immune type	147570	REa, A	Interferon, immune, deficiency (1)	10(Ifg)
12q24.1	PAH, PKU1	C	Phenylalanine hydroxylase	261600	REa, A, Fd	Phenylketonuria (3); [Hyperphenylalaninemia, mild] (3)	10(Pah)
12q24.2	ALDH2	C	Aldehyde dehydrogenase-2, mitochondrial	100650	REa, A, H	Alcohol intolerance, acute (1); ?Fetal alcohol syndrome (1)	4(Aldh-2)
Chr.12	DGU	P	DNA glycosylase, uracil	191525	REa	?Bloom syndrome, 210900 (2)	
Chr.12	MVLK	P	Mevalonate kinase	251170	REa	Mevalonicaciduria (1)	
13q12.2-q13	MBS, MOBS	L	Moebius syndrome	157900	Ch	?Moebius syndrome (2)	
13q14-q21	WND, WD	C	Wilson disease	277900	F, Fd	Wilson disease (2)	
13q14-q31	LSD	L	Letterer-Siwe disease	246400	Ch	?Letterer-Siwe disease (2)	
13q14.1	OSRC	P	Osteosarcoma	259500	Ch	Osteosarcoma, retinoblastoma-related (2)	
13q14.1-q14.2	RB1	C	Retinoblastoma-1	180200	Ch, F, Fd	Retinoblastoma (2)	14(Rb-1)
13q22.1-q32.1	HSCR, MGC	P	Megacolon (Hirschsprung disease)	249200	Ch	Megacolon (2)	
13q32	PCCA	C	Propionyl CoA carboxylase, alpha polypeptide	232000	REa, D	Propionicacidemia, type I or pccA type (1)	
13q34	DJS	L	Dubin-Johnson syndrome	237500	LD	?Dubin-Johnson syndrome (2)	
13q34	F7	C	Coagulation factor VII	227500	D	Factor VII deficiency (1)	
13q34	F10	C	Coagulation factor X	227600	D, A, REa	Factor X deficiency (1)	
13q34	HHHS	L	Hyperornithinemia-hyperammonemia-homocitrullinemia syndrome	238970	D	?HHH syndrome (2)	

Table 1—contd

Location	Symbol	Status	Title	MIM#	Method	Disorder	Mouse
Chr.13	BRCD1, DBC, BCDS1	P	Breast cancer, ductal, suppressor-1	211410	D	Breast cancer, ductal (2)	
Chr.13	UVDR, ERCM2	P	UV-damage, excision repair of (XP complementation group I)	192060	S	?Xeroderma pigmentosum, one type (1)	
14q	USH1A, USH1	P	Usher syndrome-1A (autosomal recessive, severe)	276900	Fd	Usher syndrome, type 1A (2)	
14q11.2	TCRA	C	T-cell antigen receptor, alpha polypeptide	186880	H, REa, A	Leukemia/lymphoma, T-cell (3)	14(Tcra)
14q12	MYH7, MYHCB	P	Myosin, heavy polypeptide-7, cardiac muscle, beta	160760	REa, RE, D, A	Hypertrophic cardiomyopathy, one form, 192600 (3)	
14q13.1	NP, NP1	C	Nucleoside phosphorylase	164050	S, D	Nucleoside phosphorylase deficiency, immunodeficiency due to (1)	14(Np-1,2)
14q21-q31	GALC	P	Galactocerebrosidase	245200	REa, A, H	Krabbe disease (1)	12(tw)
14q22	CSNU	L	Cystinuria	220100	Ch	?Cystinuria (2)	
14q22-q23.2	SPTB, SPH1	C	Spectrin, beta, erythrocytic	182870	REb, F, H, REa, A, RE	Elliptocytosis-3 (2); Spherocytosis-1 (3)	12(Sptb-1)
14q23-q24.2	HOS	I	Holt-Oram syndrome	142900	Ch	?Holt-Oram syndrome (2)	
14q31	TSHR	C	Thyroid-stimulating hormone receptor	275200	REa, Fd, A	Hypothyroidism, nongoitrous, due to TSH resistance (1)	
14q31-q32.1	CBG	P	Corticosteroid binding globulin	122500	A	[Transcortin deficiency] (1)	
14q32	CKBE	P	Creatine kinase, brain type, ectopic expression of	123270	F	[Creatine kinase, brain type, ectopic expression of] (2)	
14q32	SIV	L	Situs inversus viscerum	270100	H	?Situs inversus viscerum (2)	
14q32	VP, PPOX	P	Porphyria variegata (protoporphyrinogen oxidase)	176200	F	Porphyria variegata (2)	
14q32.1	AACT	C	Alpha-1-antichymotrypsin	107280	REa, A, Fd, REn	Alpha-1-antichymotrypsin deficiency (1)	
14q32.1	PI, AAT	C	Protease inhibitor (alpha-1-antitrypsin)	107400	F, S, A, D, EM, Fd	Emphysema-cirrhosis (1); Hemorrhagic diathesis due to 'antithrombin' Pittsburgh (1); Emphysema (1)	12(Aat)
14q32.1	TCL1	C	T-cell lymphoma-1	186960	Ch, RE	Leukemia/lymphoma, T-cell (2)	
14q32.33	IGH	C	IMMUNOGLOBULIN HEAVY CHAIN GENE CLUSTER		REa, A	?Combined variable hypogammaglobulinemia (1)	12(Igh)
14q32.33	IGHR	L	Immunoglobulin heavy chain regulator	144120	F	?Hyperimmunoglobulin G1 syndrome (2)	
Chr.14	MPS3C	L	Sanfilippo disease, type IIIC	252930	Ch	?Sanfilippo disease, type IIIC (2)	
Chr.14	PYGL, PPYL	P	Phosphorylase, glycogen, liver	232700	REb	Hers disease, or glycogen storage disease VI (1)	12(Pygl)
Chr.14	RMCH	P	Rod monochromacy	216900	Ch	Rod monochromacy (2)	
15q11	DLX1	L	Dyslexia-1	127700	Fc, Fd	?Dyslexia-1 (2)	
15q11	PWCR, PWS	C	Prader-Willi syndrome	176270	Ch, D	Prader-Willi syndrome (2)	
15q11-q13	ANCR, AGMS	C	Angelman syndrome	234400	Ch, D	Angelman syndrome (2)	
15q11-q13	ITO	L	Hypomelanosis of Ito	146150	Ch	?Hypomelanosis of Ito (2)	
15q14-q15	IVD	P	Isovaleryl CoA dehydrogenase	243500	REa	Isovalericacidemia (1)	
15q15	EPB42	C	Erythrocyte surface protein band 4.2	177070	A	Spherocytosis, hereditary, Japanese type (1)	
15q15-q22	LGMD2	C	Limb-girdle muscular dystrophy, autosomal recessive	253600	Fd	Muscular dystrophy, limb-girdle, autosomal recessive (2)	
15q21-q22	B2M	C	Beta-2-microglobulin	109700	S, D, H	Hemodialysis-related amyloidosis (1)	2(B2m)
15q21-q23	LIPC, LIPH, HL, HTGL	C	Hepatic triglyceride lipase	151670	REa, A	?Hepatic lipase deficiency (1)	9(Hl)
15q21.1	CYP19, ARO	C	Cytochrome P450, subfamily XIX (aromatization of androgens)	107910	REa, A, H	?Gynecomastia, familial, due to increased aromatase activity (1)	9(Cyp19)
15q21.1	FBN1, FBN, MFS1	C	Fibrillin-1	134797	A, Fd	Marfan syndrome, 154700 (3)	
15q22	PML, MYL	P	Acute promyelocytic leukemia, inducer of	102578	Ch, RE	Leukemia, acute promyelocytic (2)	
15q23-q24	HEXA, TSD	C	Hexosaminidase A (alpha polypeptide)	272800	S, D, A	Tay-Sachs disease (1); GM2-gangliosidosis, juvenile, adult (1); [Hex A pseudodeficiency] (1)	9(Hexa)
15q23-q25	ETFA, GA2	P	Electron transfer flavoprotein, alpha polypeptide	231680	REa, A	Glutaricaciduria, type II (1)	
15q23-q25	FAH	C	Fumarylacetoacetase	276700	A, REa	Tyrosinemia, type I (1)	
15q24-q25	CTSH	P	Cathepsin H	116820	REa, A	?Batten disease, one form, 204200 (1)	
Chr.15	XPF	P	Xeroderma pigmentosum, group F	278760	M	Xeroderma pigmentosum, type F (2)	
16pter-p13.3	HBA1	C	Hemoglobin alpha-1	141800	HS	Thalassemias, alpha- (1); Methemoglobinemias, alpha- (1); Erythremias, alpha- (1); Heinz body anemias, alpha- (1)	11(Hba)
16pter-p13.3	HBHR	P	Hb H mental retardation syndrome	141750	F	Hb H mental retardation syndrome (2)	
16p13.31-p13.12	PKD1, APKD	C	Adult polycystic kidney disease	173900	F, Fd	Polycystic kidney disease (2)	?17(Pkd-1)
16p13.3	RSTS, RTS	C	Rubinstein-Taybi syndrome	180849	Ch	Rubinstein-Taybi syndrome (2)	
16p13	HAGH, GLO2	C	Hydroxyacyl glutathione hydrolase; glyoxalase II	138760	S	[Glyoxalase II deficiency] (1)	
16p13	MEF, FMF	P	Familial Mediterranean fever	249100	Fd	Familial Mediterranean fever (2)	
16p13	TSC4	P	Tuberous sclerosis-4	191092	Fd	Tuberous sclerosis-4 (2)	
16p12	CLN3, BTS	C	Ceroid-lipofuscinosis, neuronal-3, juvenile (Batten disease)	204200	F, Fd	Batten disease (2)	

Table 1—contd

Location	Symbol	Status	Title	MIM#	Method	Disorder	Mouse
16q12-q13.1	PHKB	C	Phosphorylase kinase, beta polypeptide	172490	REa, A	?Phosphorylase kinase deficiency of liver and muscle, 261750 (2)	
16q13-q22.1	CES1, SES1	P	Carboxyesterase-1	114835	REa	?Monocyte carboxyesterase deficiency (1)	8(Ces-1)
16q21	CETP	P	Cholesteryl ester transfer protein, plasma	118470	REa, A	[CETP deficiency] (1)	
16q22-q24	ALDOA, ALDA	C	Aldolase A, fructose-bisphosphatase	103850	REa, REb, A	?Aldolase A deficiency (1)	
16q22.1	CTM, CPM, CAM	C	Cataract, Marner type	116800	F	Cataract, Marner type (2)	
16q22.1	LCAT	C	Lecithin-cholesterol acyltransferase	245900	F, LD, A, REa	Norum disease (3); Fish-eye disease (3)	8(Lcat)
16q22.1-q22.3	TAT	C	Tyrosine aminotransferase, cytosolic	276600	REa, A, H, D	Tyrosinemia, type II (1)	8(Tat)
16q24	APRT	C	Adenine phosphoribosyltransferase	102600	S, D	Urolithiasis, 2,8-dihydroxyadenine (1)	8(Aprt)
16q24	CYBA	C	Cytochrome b-245, alpha polypeptide	233690	REa, A	Chronic granulomatous disease, autosomal, due to deficiency of CYBA (3)	
Chr.16	ATP2A1, ATP2A	P	ATPase, Ca++ transporting, fast-twitch, 1	108730	REa	Brody myopathy (1)	
Chr.16	CTH	P	Cystathionase	219500	S	[Cystathioninuria] (1)	
17pter-p12	GP1BA	P	Glycoprotein Ib, platelet, alpha polypeptide	231200	A	Bernard-Soulier syndrome (1)	
17pter-p12	PLI	P	Alpha-2-plasmin inhibitor	262850	Psh	Plasmin inhibitor deficiency (1)	
17p13.3	BCPR	L	Breast cancer-related regulator of TP53	113721	D	?Breast cancer (1)	
17p13.3	MDCR, MDLS, MDS	C	Miller-Dieker lissencephaly syndrome	247200	Ch, D	Miller-Dieker lissencephaly syndrome (2)	11(Mds)
17p13.1	TP53, CRCR2, CRC17	C	Tumor protein p53	191170	REa, A, D	Colorectal cancer, 114500 (3); Li-Fraumeni syndrome (1)	11(Trp53)
17p11.2	CMT1A	C	Charcot-Marie-Tooth disease, slow nerve conduction type Ia	118220	Fd, D, A	Charcot-Marie-Tooth neuropathy, slow nerve conduction type Ia (2)	11(Tr)
17p11.2	SMCR	C	Smith-Magenis syndrome chromosome region	182290	Ch	Smith-Magenis syndrome (2)	
17q11-q12	EDH17B1, EDHB17A	C	Estradiol 17-beta-dehydrogenase-1	264300	A, REa	Pseudohermaphroditism, male, with gynecomastia (1); Polycystic ovarian disease (1)	
17q11.2	NF1, VRNF	C	Neurofibromatosis, von Recklinghausen type	162200	Fd, EM, Ch, F	Neurofibromatosis, von Recklinghausen (2)	
17q11.2	WSS	P	Watson syndrome	193520	Fd	Watson syndrome (2)	
17q12-q21	KRT14, K14	P	Keratin-14	148066	REa	Epidermolysis bullosa simplex, 131900 (3); Epidermolysis bullosa simplex, Dowling-Meara type, 131760 (3)	
17q21	ACAC, ACC	P	Acetyl-CoA carboxylase	200350	A	Acetyl-CoA carboxylase deficiency (1)	
17q21	BRCA1	P	Breast cancer-1, early onset	113705	Fd	Breast cancer-1, early onset (2)	
17q21-q22	EPB3, EMPB3	C	Erythroid membrane protein band 3	109270	REa, RE, Fd	[Acanthocytosis, one form] (1); [Elliptocytosis, Malaysian-Melanesian type] (1)	
17q21-q22	GALK	C	Galactokinase	230200	S, Ch, R, C	Galactokinase deficiency (1)	11(Glk)
17q21-q22	KRT10	C	Keratin-10	148080	REa, A, REn	Epidermolytic hyperkeratosis, 113800 (1)	
17q21.1	RARA	C	Retinoic acid receptor, alpha polypeptide	180240	A, Ch	Leukemia, acute promyelocytic (1)	11(Rara)
17q21.3-q22	MPO	C	Myeloperoxidase	254600	REa, A, F, Ch, C	Myeloperoxidase deficiency (1)	11(Mpo)
17q21.31-q22.05	COL1A1	C	Collagen I, alpha-1 polypeptide	120150	C, M, A, REa	Osteogenesis imperfecta, 2 or more clinical forms (3); Ehlers-Danlos syndrome, type VIIA1 (3)	11(Cola-1)
17q21.32	ITGA2B, GP2B, CD41B	C	Integrin, alpha IIB (platelet glycoprotein IIb of IIb/IIIa complex, antigen CD41B)	273800	A, REb, REa, RE, F, LD	Glanzmann thrombasthenia, type A (1)	
17q21.32	ITGB3, GP3A	C	Integrin, beta-3 (platelet glycoprotein IIIa; antigen CD61)	173470	REa, REb, A, RE, F, LD	Glanzmann thrombasthenia, type B (1)	
17q22-q24	CSH1, CSA, PL	C	Chorionic somatomammotropin A	150200	REa, A	[Placental lactogen deficiency] (1)	13(Pl-1)
17q22-q24	GH1, GHN	C	Growth hormone, normal	139250	REa, A, Fd	Isolated growth hormone deficiency, Illig type with absent GH and Kowarski type with bioinactive GH (3)	11(Gh)
17q23	GAA	C	Acid alpha-glucosidase	232300	S, A, D, C	Pompe disease (1); Acid-maltase deficiency, adult (1)	
17q23-qter	APOH	C	Apolipoprotein H (beta-2-glycoprotein I)	138700	Fd, REa	[Apolipoprotein H deficiency] (1)	11(Apoh)
17q23.1-q25.3	SCN4A, HYPP, NAC1A	C	Sodium channel, voltage-gated, type 4, alpha polypeptide	170500	REa, Fd	Hyperkalemic periodic paralysis (3); Paramyotonia congenita (3); Myotonia congenita, atypical acetazolamide-responsive (3)	
18p11.32	MCL	L	Multiple hereditary cutaneous leiomyomata	150800	Ch	?Leiomyomata, multiple hereditary cutaneous (2)	
18q11-q12	LCFS2	L	Lynch cancer family syndrome II	114400	F	?Lynch cancer family syndrome II (2)	
18q11.2-q12.1	TTR, PALB, TBPA	C	Thyroxine-binding prealbumin (transthyretin)	176300	REa, A	Amyloid neuropathy, familial, several allelic types (1); [Dystransthyretinemic hyperthyroxinemia](1); Amyloidosis, senile systemic (1)	18(Palb)

Table 1—contd

Location	Symbol	Status	Title	MIM#	Method	Disorder	Mouse
18q21-q22.2	CORD, RCRD1	L	Retinal cone-rod dystrophy-1	120970	Ch	?Retinal cone-rod dystrophy (2)	
18q21.3	BCL2	C	B-cell CLL/lymphoma-2	151430	Ch, RE, REn	Leukemia/lymphoma, B-cell, 2 (2)	1(Bcl-2)
18q21.3	FECH, FCE	C	Ferrochelatase	177000	A, REb	Protoporphyria, erythropoietic (1)	
18q22.1	GTS	L	Gilles de la Tourette syndrome	137580	Ch	?Tourette syndrome (2)	
18q23.3	DCC, CRC18	C	Colorectal cancer-related sequence-18	120470	D, RE	Colorectal cancer (1)	18(Dcc)
Chr.18	CYB5	P	Cytochrome b5	250790	Psh	Methemoglobinemia due to cytochrome b5 deficiency (1)	
19p13.3-p13.2	AMH, MIF	P	Anti-Mullerian hormone	261550	REa, A	Persistent Mullerian duct syndrome (1)	10(Amh)
19p13.3-p13.2	ATHS, ALP	P	Atherogenic lipoprotein phenotype (atherosclerosis susceptibility)	108725	Fd	Atherosclerosis, susceptibility to (2)	
19p13.3-p13.2	C3	C	Complement component-3	120700	F, S, A, REa	C3 deficiency (1)	17(C3)
19p13.3-p13.2	INSR	C	Insulin receptor	147670	REa, A, REb	Leprechaunism (1); Diabetes mellitus, insulin-resistant, with acanthosis nigricans (1); ?Rabson-Mendenhall syndrome (1)	8(Insr)
19p13.3-p13.2	TCF3, E2A	P	Transcription factor-3 (E2A immunoglobulin enhancer binding factors E12/E47)	147141	REa, A	Leukemia, acute lymphoblastic (1)	
19p13.2-p13.1	LDLR,FHC	C	Familial hypercholesterolemia (LDL receptor)	143890	F, REa, A	Hypercholesterolemia, familial (3)	9(Ldlr)
19p13.2-q12	MANB	C	Lysosomal alpha-D-mannosidase-B	248500	S	Mannosidosis (1)	
19p13.1	RFX1	C	Regulatory factor (trans-acting) 1 (influences HLA class II expression)	209920	A	Severe combined immunodeficiency, HLA class II-negative type (1)	
19p13	LYL1	P	Leukemia, lymphoid, 1	151440	Ch	Leukemia, T-cell acute lymphoblastoid (2)	8(Lyl-1)
19cen-q12	GPI	C	Glucose phosphate isomerase; neuroleukin	172400	S, D	Hemolytic anemia due to glucosephosphate isomerase deficiency (1); Hydrops fetalis, one form (1)	7(Gpi-1)
19cen-q13.11	PEPD	C	Peptidase D (prolidase)	170100	S, F, H, Fd	Prolidase deficiency (1)	7(Pep-4)
19q12-q13.2	CCO, CCD	C	Central core disease of muscle	117000	Fd	Central core disease of muscle (2)	
19q12-q13.2	PVS	C	Polio virus sensitivity	173850	S	{Polio, susceptibility to} (2)	9(Pvs)
19q13	BCL3	C	B-cell CLL/lymphoma-3	109560	Ch, S, H	Leukemia/lymphoma, B-cell, 3 (2)	7(Bcl-3)
19q13.1	APOE	C	Apolipoprotein E	107741	F, REa, LD, A, Fd	Hyperlipoproteinemia, type III (1)	7(Apoe)
19q13.1	APOC2	C	Apolipoprotein C-II	207750	REa, F, LD, A, Fd	Hyperlipoproteinemia, type Ib (1)	
19q13.1	RYR1, RYDR, MHS	C	Ryanodine receptor (sarcoplasmic reticulum calcium release channel)	180901	A, Fd, H	Malignant hyperthermia, 145600 (3)	7(Ryr)
19q13.1-q13.2	BCKDHA, MSUD1	C	Branched-chain keto acid dehydrogenase E1, alpha polypeptide	248600	REa, REb, A	Maple syrup urine disease, type 1 (3)	
19q13.2-q13.3	DM	C	Myotonic dystrophy	160900	F, Fd	Myotonic dystrophy (2)	
19q13.2-q13.3	ERCC2, EM9	C	Excision repair cross complementing rodent repair deficiency, complementation group-2	126340	S, RE	Xeroderma pigmentosum, group D, 278730 (1)	7(Ercc-2)
19q13.3	LIG1	C	Ligase I, DNA, ATP-dependent	126391	REa, A	DNA ligase I deficiency (1)	
19q13.32	LHB	C	Luteinizing hormone, beta polypeptide	152780	RE	Hypogonadism, hypergonadotropic (1); ?Male pseudohermaphroditism due to defective LH (1)	7(Lhb)
Chr.19	BCT2	P	Branched-chain aminotransferase-2	113530	S	?Hypervalinemia or hyperleucine-isoleucinemia (1)	
Chr.19	ETFB	P	Electron transfer flavoprotein, beta polypeptide	130410	REa	Glutaricaciduria, type IIc (1)	
20pter-p12.21	ARVP, VP	P	Arginine vasopressin-neurophysin II	192340	REa, RE, Fd	Diabetes insipidus, neurohypophyseal, 125700 (1)	
20pter-p12	PRNP, PRIP	C	Prion protein (p27-30)	176640	REa, REb, A	Creutzfeldt-Jakob disease, 123400 (3); Gerstmann-Straussler disease, 137440 (3); Insomnia, fatal familial (3)	2(Prn-p)
20p13	HOS	I	Holt-Oram syndrome	142900	Ch	?Holt-Oram syndrome (2)	
20p11.23-qter	GHRF	C	Growth hormone releasing factor; somatocrinin	139190	REa, REb, Ch	?Isolated growth hormone deficiency due to defect in GHRF (1)	
20p11.2	AGS, AHD	C	Alagille syndrome (arteriohepatic dysplasia)	118450	Ch, D	Alagille syndrome (2)	
20p11	CST3	C	Cystatin C	105150	REa, A	Cerebral amyloid angiopathy (1)	
20p	ITPA	C	Inosine triphosphatase-A	147520	S	[Inosine triphosphatase deficiency] (1)	2(Itp)
20q13	MODY1	C	Maturity-onset diabetes of the young, type I	125850	Fd	MODY, type I (2)	
20q13.1	PPGB, GSL, NGBE, GLB2	P	Protective protein for beta-galactosidase	256540	S, A	Galactosialidosis (1)	
20q13.11	ADA	C	Adenosine deaminase	102700	S, D, REa, F, A	Severe combined immunodeficiency due to ADA deficiency (1); Hemolytic anemia due to ADA excess (1)	2(Ada)
20q13.2	GNAS1, GNAS, GPSA	C	Guanine nucleotide-binding protein (G protein), alpha-stimulating activity polypeptide-1	139320	REa, H, A, Fd	Pseudohypoparathyroidism, type Ia (1); McCune-Albright polyostotic fibrous dysplasia, 174800 (1); Pituitary tumor, growth-hormone-secreting (1)	2(Gnas)

Table 1—contd

Location	Symbol	Status	Title	MIM#	Method	Disorder	Mouse
10q13.2-q13.3	EBN, BNS	C	Epilepsy, benign neonatal	121200	Fd	Epilepsy, benign neonatal (2)	
20q13.2-q13.3	FA, FA1, FACA	P	Fanconi anemia-1	227650	Fd	Fanconi anemia-1 (2)	
Chr.20	BMP2, BMP2A	P	Bone morphogenetic protein-2a	112261	H, REa	?Fibrodysplasia ossificans progressiva (1)	2(Bmp2a)
21q11.2	MST	L	Myeloproliferative syndrome, transient	159595	Ch	?Leukemia, transient (2)	
21q21.3-q22.05	APP, AAA, CVAP	C	Amyloid beta (A4) precursor protein	104760	REa, A, Fd, RE	Amyloidosis, cerebroarterial, Dutch type (1); Alzheimer disease, APP related (3)	16(App)
21q22	AML1	P	aml-1 (acute myeloid leukemia) oncogene	151385	Ch	Leukemia, acute myeloid (2)	
21q22.1-q22.2	ALS1	P	Amyotrophic lateral sclerosis-1	105400	Fd	Amytrophic lateral sclerosis, one form (2)	
21q22.3	CBS	C	Cystathionine beta-synthase	236200	S, D, A	Homocystinuria, B6-responsive and nonresponsive types (1)	17(Cbs)
21q22.3	EPM1	P	Epilepsy, progressive myoclonic 1	254800	Fd	Epilepsy, progressive myoclonus (2)	
21q22.3	ITGB2, CD18, LCAMB, LAD	C	Integrin, beta-2 (antigen CD18 (p95), lymphocyte function-associated antigen-1; macrophage antigen, beta polypeptide)	116920	S, A, Fd	Leukocyte adhesion deficiency (1)	7(Ly-15)
21q22.3	PFKL	C	Phosphofructokinase, liver type	171860	S, D, Fd	Hemolytic anemia due to phosphofructokinase deficiency (1)	17(Pfkl)
22q11	CECR, CES	C	Cat eye syndrome	115470	Ch, A, D	Cat eye syndrome (2)	
22q11	DGCR, DGS	C	DiGeorge syndrome	188400	Ch, D	DiGeorge syndrome (2)	
22q11	HCF2, HC2	C	Heparin cofactor II	142360	REb, REa	Thrombophilia due to heparin cofactor II deficiency (1)	
22q11	NAGA	C	Acetylgalactosaminidase, alpha-N-(alpha-galactosidase B)	104170	S, Ch	Alpha-NAGA deficiency (1)	
22q11	VCFS	L	Velocardiofacial syndrome	192430	D	?Velocardiofacial syndrome (2)	
22q11.1-q11.2	GGT2, GTG	C	Gamma-glutamyltranspeptidase-2	231950	A, S, F, RE	Glutathioninuria (1)	
22q11.2-q12.2	CYP2D, P450C2D	C	Cytochrome P450, subfamily IID	124030	F	{?Parkinsonism, susceptibility to} (1); Debrisoquine sensitivity (1)	15(Cyp2d)
22q11.2-qter	SGLT1, NAGT	P	Sodium-glucose transporter-1	182380	REa	Glucose/galactose malabsorption (1)	
22q11.2-qter	TCN2, TC2	C	Transcobalamin II	275350	F, S, D	Transcobalamin II deficiency (1)	11(Tcn-2)
22q11.21	BCR, CML, PHL	C	Chronic myeloid leukemia; breakpoint cluster region	151410	Ch, RE	Leukemia, chronic myeloid (3)	
22q11.21-q13.1	NF2, ACN	C	Acoustic neuroma	101000	RE, F	Acoustic neuroma (2)	
22q12	ES	C	Ewing sarcoma (neuroepithelioma)	133450	Ch	Ewing sarcoma (2); Neuroepithelioma (2)	
22q12.3-q13.1	PDGFB, SIS	C	Platelet-derived growth factor, beta polypeptide (oncogene SIS)	190040	REa, Fd	Meningioma (3)	15(Pdgfb)
22q12.3-qter	MGCR, MGM	C	Meningioma	156100	Ch, RE, D	Meningioma (2)	
22q13-qter	ACR, ACRS	P	Acrosin (proacrosin)	102480	REa	?Male infertility due to acrosin deficiency (2)	15(Acr)
22q13.1	ADSL, ADS	P	Adenylosuccinase (adenylosuccinate lyase)	103050	S, REa	Adenylosuccinase deficiency (1)	
22q13.31-qter	ARSA	C	Arylsulfatase A	250100	S, D	Metachromatic leukodystrophy (1)	15(As-2)
22q13.31-qter	DIA1	C	NADH-diaphorase-1 (cytochrome b5 reductase)	250800	S, REa	Methemoglobinemia, enzymopathic (1)	15(Dia-1)
Xpter-p21	CSF2RA	P	Colony-stimulating factor-2 receptor, alpha, low-affinity (granulocyte-macrophage)	306250	A	Leukemia, acute myeloid, M2 type (1)	
Xp22.32	STS, ARSC1, SSDD	C	Steroid sulfatase, microsomal	308100	F, S, D	Ichthyosis, X-linked (3); Placental steroid sulfatase deficiency (3)	X,Y(Sts)
Xp22.31	DHOF, FODH	P	Focal dermal hypoplasia	305600	Ch	Focal dermal hypoplasia (2)	
Xp22.3	CDPX1, CDPXR	C	Chondrodysplasia punctata, X-linked recessive	302950	D, Fd	Chondrodysplasia punctata, X-linked recessive (2)	
Xp22.3	KAL, KMS, ADMLX	C	Kallmann syndrome	308700	F, Fd, D, REa, REn	Kallmann syndrome (2)	
Xp22.3	OA1	C	Ocular albinism, Nettleship-Falls type	300500	F, Fd	Ocular albinism, Nettleship-Falls type (2)	
Xp22.3-p22.1	AMELX, AMG, AIH1, AMGX	C	Amelogenin (amelogenesis imperfecta, hypoplastic type I)	301200	REa, A, Fd	Amelogenesis imperfecta (1)	X(Amel)
Xp22.3-p21.1	NHS	C	Nance-Horan cataract-dental syndrome	302350	Fd	Nance-Horan syndrome (2)	?X(Xcat)
Xp22.3-p21.1	POLA	C	Polymerase, DNA directed, alpha	312040	S	?N syndrome, 310465 (1)	X(Pola)
Xp22.3-p22.1	RS	C	Retinoschisis	312700	F, Fd	Retinoschisis (2)	
Xp22.2	CMTX2	P	Charcot-Marie-Tooth disease, X-linked-2, recessive	302801	Fd	Charcot-Marie-Tooth neuropathy, X-linked-2, recessive (2)	
Xp22.2-p21.3	ZFX	C	Zinc finger protein, X-linked	314980	Ch, REa, Fd	46,XY female (2)	X(Zfx)
Xp22.2-p21.2	KFSD	P	Keratosis follicularis spinulosa decalvans	308800	Fd	Keratosis follicularis spinulosa decalvans (2)	
Xp22.2-p22.1	CLS	P	Coffin-Lowry syndrome	303600	Fd	Coffin-Lowry syndrome (2)	
Xp22.2-p22.1	HYP, HPDR1	C	Hereditary hypophosphatemia	307800	Fd	Hypophosphatemia, hereditary (2)	X(Hyp)
Xp22.2-p22.1	MRXS1	P	Mental retardation, X-linked, syndromic-1, with dystonic movements, ataxia, and seizures	309510	Fd	Mental retardation, X-linked, syndromic-1, with dystonic movements, ataxia, and seizures (2)	

Table 1—contd

Location	Symbol	Status	Title	MIM#	Method	Disorder	Mouse
Xp22.2-p22.1	PDHA1, PHE1A	C	Pyruvate dehydrogenase, E1-alpha polypeptide-1	312170	REa, A	Pyruvate dehydrogenase deficiency (1)	X(Pdha-1)
Xp22.2-p22.1	PHK, PHKA2	P	Phosphorylase kinase deficiency, liver (glycogen storage disease type VIII)	306000	Fd, REa	Glycogen storage disease, X-linked hepatic (2)	
Xp22	AGMX2, XLA2, IMD6	P	Agammaglobulinemia, X-linked 2 (with growth hormone deficiency)	300310	Fd	Agammaglobulinemia, type 2, X-linked (2)	X(Xid)
Xp22	AIC	C	Aicardi syndrome	304050	X/A, Ch	Aicardi syndrome (2)	
Xp22	GY	L	Hereditary hypophosphatemia II (gyro equivalent)	307810	H	?Hypophosphatemia with deafness (2)	X(Gy)
Xp22	HOMG, HSH, HMGX	L	Hypomagnesemia, X-linked primary	307600	X/A	Hypomagnesemia, X-linked primary (2)	
Xp22	MRX1	C	Mental retardation, X-linked-1, non-dysmorphic	309530	F, Fd, D	Mental retardation, X-linked-1, non-dysmorphic (2)	
Xp22	SEDL, SEDT	C	Spondyloepiphyseal dysplasia tarda	313400	Fd	Spondyloepiphyseal dysplasia tarda (2)	
Xp22-p21	GDXY, TDFX	P	Gonadal dysgenesis, XY female type	306100	F, Ch	Gonadal dysgenesis, XY female type (2)	
Xp21.3-p21.1	RP3	C	Retinitis pigmentosa-3 (RP with metallic sheen in heterozygotes)	312610	Fd, D	Retinitis pigmentosa-3 (2)	
Xp21.3-p21.2	AHC, AHX	C	Primary adrenal hypoplasia	300200	D, Fd	Adrenal hypoplasia, primary (2)	
Xp21.3-p21.2	GK	C	Glycerol kinase	307030	D, Fd	Glycerol kinase deficiency (2)	
Xp21.2	DMD, BMD	C	Dystrophin (muscular dystrophy, Duchenne and Becker types)	310200	X/A, Fd, D	Duchenne muscular dystrophy (3); Becker muscular dystrophy (3); Cardiomyopathy, dilated, X-linked (3)	X(Dmd)
Xp21.2-p21.1	XK	C	Xk blood group	314850	F, D	[McLeod phenotype] (2)	
Xp21.1	CYBB, CGD	C	Chronic granulomatous disease	306400	F, D	Chronic granulomatous disease, X-linked (3)	X(Cybb)
Xp21.1	OTC	C	Ornithine transcarbamylase	311250	L, REa, A, D	Ornithine transcarbamylase deficiency (3)	X(spf; Otc)
Xp21.1-p11.3	COD1, PCDX	P	Progressive cone dystrophy, X-linked	304020	Fd	Progressive cone dystrophy (2)	
Xp21.1-q22	MRXS6	P	Mental retardation, X-linked, syndromic-6, with gynecomastia and obesity	309585	Fd	Mental retardation, X-linked, syndromic-6, with gynecomastia and obesity (2)	
Xp21	GTD	L	Gonadotropin deficiency	306190	D	?Gonadotropin deficiency (2); ?Cryptorchidism (2)	
Xp21-p11	THC	P	Thrombocytopenia, X-linked	313900	Fd	Thrombocytopenia, X-linked (2)	
Xp11.4	NDP, ND	C	Norrie disease	310600	Fd, D	Norrie disease (2)	
Xp11.4-p11.23	PFC, PFD	C	Properdin P factor, complement (properdin P deficiency)	312060	Fd, REa, A	Properdin deficiency, X-linked (3)	X(Pfc)
Xp11.3	CSNB1	I	Congenital stationary nightblindness	310500	Fd	Nightblindness, congenital stationary, type I (2)	
Xp11.3	RP2	C	Retinitis pigmentosa-2	312600	Fd	Retinitis pigmentosa-2 (2)	
Xp11.3-p11.2	WAS, IMD2	C	Wiskott-Aldrich syndrome	301000	Fd, X/A	Wiskott-Aldrich syndrome (2)	
Xp11.23	GF1, ERYF1, NFE1	C	Globin transcription factor-1	305371	REa, A	?Hereditary persistence of fetal hemoglobin, Swiss type (2)	X(Gf-1)
Xp11.21	ALAS2, ASB, ANH1	C	Aminolevulinate, delta-, synthase-2	301300	Ch, REa, A	Anemia, sideroblastic/hypochromic (3)	
Xp11.21	IP1, IP	C	Incontinentia pigmenti-1, sporadic type	308300	X/A	Incontinentia pigmenti, sporadic type (2)	X(Td)
Xp11.2	SSRC	P	Sarcoma, synovial	312820	Ch, RE	Sarcoma, synovial (2)	
Xp11-q11	AIED, OA2	C	Aland island eye disease (ocular albinism, Forsius-Eriksson type)	300600	F, D, Fd	Ocular albinism, Forsius-Eriksson type (2)	
Xp11-q21	MRXS2	P	Mental retardation, X-linked, syndromic-2, with dysmorphism and cerebral atrophy; Prieto syndrome	309610	Fd	Mental retardation, X-linked, syndromic-2, with dysmorphism and cerebral atrophy (2)	
Xp11-q21.3	MRXS3	P	Mental retardation, X-linked, syndromic-3, with spastic diplegia	309470	Fd	Mental retardation, X-linked, syndromic-3, with spastic diplegia (2)	
Xp	CCT	L	Cataracts, congenital total	302200	Fd	?Cataract, congenital total (2)	
Xp	RTS	L	Rett syndrome	312750	Ch	?Rett syndrome (2)	
Xcen-q21.3	SGB	L	Simpson-Galabi-Behmel syndrome	312870	Fd	?Simpson-Galabi-Behmel syndrome (2)	
Xcen-q22	AR, DHTR, TFM, SBMA, KD, SMAX1	C	Androgen receptor (testicular feminization)	313700	S, Fd, REa, A	Testicular feminization (1); Reifenstein syndrome (1); Infertile male syndrome (1); Spinal and bulbar muscular atrophy of Kennedy, 313200 (3)	X(Tfm)
Xq11-q12	MRX2	L	Mental retardation, X-linked-2, non-dysmorphic	309540	Fd	Mental retardation, X-linked-2, non-dysmorphic (2)	
Xq12-q13	MNK, MK	C	Menkes disease	309400	Fc, X/A, H	Menkes disease (2)	X(Mo)
Xq12-q13	PHKA1	P	Phosphorylase kinase, muscle, alpha polypeptide	311870	REa, A	?Muscle glycogenosis (1)	X(Phka)
Xq12-q21.1	DYT3	P	Torsion dystonia-parkinsonism, Filipino type	314250	Fd	Torsion dystonia-parkinsonism, Filipino type (2)	
Xq12.2-13.1	EDA, HED	C	Anhidrotic ectodermal dysplasia	305100	X/A, H, Fd	Anhidrotic ectodermal dysplasia (2)	X(Ta)
Xq13	CMTX1, CMT2	C	Charcot-Marie-Tooth disease, X-linked-1, dominant	302800	Fd	Charcot-Marie-Tooth neuropathy, X-linked-1, dominant (2)	
Xq13	FGDY, AAS	P	Aarskog-Scott syndrome (faciogenital dysplasia)	305400	X/A	Aarskog-Scott syndrome (2)	
Xq13	PGK1, PGKA	C	Phosphoglycerate kinase-1	311800	S, R, REb, Fd	Hemolytic anemia due to PGK deficiency (1); Myoglobinuria/ hemolysis due to PGK deficiency (1)	X(Pgk-1)

Table 1—contd

Location	Symbol	Status	Title	MIM#	Method	Disorder	Mouse
Xq13-q21	WWS	P	Wieacker-Wolff syndrome	314580	Fd	Wieacker-Wolff syndrome (2)	
Xq13-q21.1	DFN3	C	Conductive deafness with stapes fixation	304400	Fd, D	Deafness, conductive, with stapes fixation (2)	
Xq13-q21.31	CPX	C	Cleft palate, X-linked	303400	Fd, D	Cleft palate, X-linked (2)	
Xq13-q22	MRXS4	P	Mental retardation, X-linked, syndromic-4, with congenital contractures and low fingertip arches	309605	Fd	Mental retardation, X-linked, syndromic-4, with congenital contractures and low fingertip arches (2)	
Xq13.1	RPS4X, CCG2, SCAR	C	Ribosomal protein S4, X-linked	312760	A, REa	Turner syndrome (1)	X(Rps4x)
Xq13.1-q21.1	SCIDX1, SCIDX, IMD4	C	Severe combined immunodeficiency, X-linked	300400	Fd	Severe combined immunodeficiency, X-linked (2)	
Xq21	AHDS, AHS	P	Allan-Herndon mental retardation syndrome	309600	Fd	Allan-Herndon syndrome (2)	
Xq21-q22	SPG2, SPPX2	P	Spastic paraplegia, X-linked, uncomplicated	312920	Fd	Spastic paraplegia, X-linked, uncomplicated (2)	
Xq21-q22	TBG	P	Thyroxine-binding globulin	314200	REa, A	[Euthyroidal hyper- and hypothyroxinemia] (1)	
Xq21.2	CHM, TCD	C	Choroideremia	303100	Fd, LD, D, A, Ch, X/A	Choroideremia (2)	
Xq21.3-q22	AGMX1, IMD1, XLA	C	Agammaglobulinemia, X-linked 1 (Bruton)	300300	H, Fd	Agammaglobulinemia, type 1, X-linked (2)	
Xq21.3-q22	MGC1, MGCN	P	Megalocornea, X-linked	309300	Fd	Megalocornea, X-linked (2)	
Xq21.3-q22	PHP, GHDX	L	Growth hormone deficiency, X-linked (panhypopituitarism, X-linked)	312000	Fd	?Growth hormone deficiency, X-linked (2)	
Xq22	COL4A5, ATS, ASLN	C	Collagen IV, alpha-5 polypeptide	303630	REa, A, Fd	Alport syndrome, 301050 (3)	
Xq22	GLA	C	Galactosidase, alpha	301500	S, R, A, Fd	Fabry disease (3)	X(Ags)
Xq22	PLP, PMD	C	Proteolipid protein; Pelizaeus-Merzbacher disease	312080	REa, A, Ch, R, Fd	Pelizaeus-Merzbacher disease (3)	X(Plp(jp))
Xq22-q24	PRPS1	C	Phosphoribosylpyrophosphate synthetase I	311850	S, R, REa, A	Phosphoribosylpyrophosphate synthetase-related gout (1)	
Xq24-q27	HIGM1, IMD3	P	X-linked immunodeficiency with hyper-IgM	308230	Fd	Immunodeficiency, X-linked, with hyper-IgM (2)	
Xq25	LYP, IMD5, XLP, XLPD	C	Lymphoproliferative syndrome, X-linked	308240	Fd, D	Lymphoproliferative syndrome, X-linked (2)	
Xq25-q27	PGS, MRXS5	P	Pettigrew syndrome (mental retardation, X-linked, with Dandy-Walker malformation, basal ganglia disease, and seizures)	304340	Fd	Mental retardation, X-linked, syndromic-5, with Dandy-Walker malformation, basal ganglia disease, and seizures (2)	
Xq26-q27	BFLS	P	Borjeson-Forssman-Lehmann syndrome	301900	Fd	Borjeson-Forssman-Lehmann syndrome (2)	
Xq26-q27	HPT, HPTX, HYPX	P	Hypoparathyroidism, X-linked	307700	Fd	Hypoparathyroidism, X-linked (2)	
Xq26-q27	POF1, POF	L	Premature ovarian failure	311360	Ch	Ovarian failure, premature (2)	
Xq26-q27.2	HPRT	C	Hypoxanthine-guanine phosphoribosyltransferase	308000	S, M, C, R, REa, Fd	Lesch-Nyhan syndrome (3); HPRT-related gout (1)	X(Hprt)
Xq26.1	OCRL, LOCR	C	Lowe oculocerebrorenal syndrome	309000	X/A, Fd	Lowe syndrome (2)	
Xq26.3-q27.1	ADFN, ALDS	P	Albinism-deafness syndrome	300700	Fd	Albinism-deafness syndrome (2)	
Xq27-q28	ANOP1	L	Anophthalmos-1 (with mental retardation but without anomalies)	301590	F	?Anophthalmos-1 (2)	
Xq27-q28	IP2	P	Incontinentia pigmenti-2 (familial, male-lethal type)	308310	Fd	Incontinentia pigmenti, familial (2)	X(?Str)
Xq27.1-q27.2	F9, HEMB	C	Coagulation factor IX (hemophilia B)	306900	REa, A, Fd, D, X/A, RE	Hemophilia B (3)	X(Cf-9)
Xq27.3	FRAXA, FMR1	C	Fragile site Xq27.3	309550	Ch, F, Fd, RE	Martin-Bell syndrome (2)	X(Fmr-1)
Xq28	ALD	C	Adrenoleukodystrophy	300100	F, Fd, D	Adrenoleukodystrophy (2); Adrenomyeloneuropathy (2)	
Xq28	CBBM, BCM	C	Blue-monochromatic colorblindness (blue cone monochromacy)	303700	F, Fd, RE	Colorblindness, blue monochromatic (3)	
Xq28	CDPX2, CPXD, CPX	L	Chondrodysplasia punctata, X-linked dominant (Happle syndrome)	302960	H	Chondrodysplasia punctata, X-linked dominant (2)	X(Bpa)
Xq28	DIR, DI1, ADHR	C	Nephrogenic diabetes insipidus (vasopressin V2-receptor)	304800	Fd, S, REa	Diabetes insipidus, nephrogenic (3)	
Xq28	DKC	P	Dyskeratosis congenita	305000	Fd	Dyskeratosis congenita (2)	
Xq28	EFE2, BTHS	C	Endocardial fibroelastosis-2 (Barth syndrome; cardioskeletal myopathy with neutropenia and abnormal mitochondria)	302060	Fd	Endocardial fibroelastosis-2 (2)	
Xq28	EMD	C	Emery-Dreifuss muscular dystrophy	310300	F, Fd, H	Emery-Dreifuss muscular dystrophy (2)	
Xq28	F8C, HEMA	C	Coagulation factor VIII (hemophilia A)	306700	F, Fd, REa, A, RE	Hemophilia A (3)	X(Cf-8)
Xq28	G6PD, G6PD1	C	Glucose-6-phosphate dehydrogenase	305900	F, S, REb, RE	G6PD deficiency (3); Favism (1); Hemolytic anemia due to G6PD deficiency (1)	X(G6pd)

Table 1—contd

Location	Symbol	Status	Title	MIM#	Method	Disorder	Mouse
Xq28	GCP, CBD	C	Deutan colorblindness (green cone pigment)	303800	F, RE, A, Fd	Colorblindness, deutan (3)	X(Rsvp)
Xq28	HSAS1, HSAS, HYCX	C	Hydrocephalus, X-linked, due to stenosis of aqueduct of Sylvius	307000	Fd	Hydrocephalus, X-linked (2)	
Xq28	IDS, MPS2, SIDS	C	Hunter syndrome (sulfoiduronate sulfatase deficiency)	309900	X/A, Fd, F, RE	Mucopolysaccharidosis II (2)	X(Ids)
Xq28	MAFD2, MDX	L	Manic-depressive illness, X-linked	309200	F	?Manic-depressive illness, X-linked (2)	
Xq28	MASA	C	MASA syndrome (complicated spastic paraplegia)	303350	Fd	MASA syndrome (2)	
Xq28	MRSD, CHRS	P	Mental retardation-skeletal dysplasia	309620	Fd	Mental retardation-skeletal dysplasia (2)	
Xq28	MRX3	L	Mental retardation, X-linked-3	309541	Fd	?Mental retardation, X-linked-3 (2)	
Xq28	MTM1, MTMX	C	Myotubular myopathy, X-linked	310400	Fd	Myotubular myopathy, X-linked (2)	
Xq28	MYP1, BED	P	Myopia-1 (Bornholm eye disease)	310460	Fd	Myopia-1 (2); Bornholm eye disease (2)	
Xq28	OPD1	P	Otopalatodigital syndrome, type I	311300	Fd	Otopalatodigital syndrome, type I (2)	
Xq28	RCP, CBP	C	Protan colorblindness (red cone pigment)	303900	F, RE, A, Fd	Colorblindness, protan (3)	X(Rsvp)
Xq28	TKC, TKCR	C	Goeminne TKCR syndrome	314300	X/A	Goeminne TKCR syndrome (2)	
Xq28	WSN, BGMR	P	Waisman syndrome (basal ganglion disorder with mental retardation)	311510	Fd	Waisman parkinsonism-mental retardation syndrome (2)	

Table 2 The morbid anatomy of the human genome (by disorder).

Disorder	*Location*
Aarskog-Scott syndrome (2)	Xq13
?Abetalipoproteinemia (1)	2p24
[Acanthocytosis, one form] (1)	17q21-q22
Acatalasemia (1)	11p13
Acetyl-CoA carboxylase deficiency (1)	17q21
Acid-maltase deficiency, adult (1)	17q23
Acoustic neuroma (2)	22q11.21-q13.1
ACTH deficiency (1)	2p25
Acyl-CoA dehydrogenase, long chain, deficiency of (1)	2q34-q35
Acyl-CoA dehydrogenase, medium chain, deficiency of (1)	1p31
Acyl-CoA dehydrogenase, short chain, deficiency of (1)	12q22-qter
Adenylosuccinase deficiency (1)	22q13.1
Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency (1)	8q21
Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency (3)	6p21.3
Adrenal hyperplasia II (1)	1p13.1
Adrenal hyperplasia V (1)	Chr.10
Adrenal hypoplasia, primary (2)	Xp21.3-p21.2
Adrenocortical carcinoma (2)	11p15.5
Adrenoleukodystrophy (2)	Xq28
Adrenomyeloneuropathy (2)	Xq28
[AFP deficiency, congenital] (1)	4q11-q13
Agammaglobulinemia, type 1, X-linked (2)	Xq21.3-q22
Agammaglobulinemia, type 2, X-linked (2)	Xp22
Aicardi syndrome (2)	Xp22
Alagille syndrome (2)	20p11.2
Albinism (3)	11q14-q21
Albinism-deafness syndrome (2)	Xq26.3-q27.1
Alcohol intolerance, acute (1)	12q24.2
?Aldolase A deficiency (1)	16q22-q24
Allan-Herndon syndrome (2)	Xq21
Alpha-1-antichymotrypsin deficiency (1)	14q32.1
Alpha-NAGA deficiency (1)	22q11
Alport syndrome, 301050 (3)	Xq22
Alzheimer disease, APP related (3)	21q21.3-q22.05
Amelogenesis imperfecta (1)	Xp22.3-p22.1
[AMP deaminase deficiency, erythrocyte] (1)	1p21-p13
Amyloid neuropathy, familial, several allelic types (1)	18q11.2-q12.1
Amyloidosis, cerebroarterial, Dutch type (1)	21q21.3-q22.05
Amyloidosis, Finnish type, 105120 (1)	9q34
Amyloidosis, Iowa type, 107680.0010 (2)	11q23
{?Amyloidosis, secondary, susceptibility to} (1)	1q21-q23
Amyloidosis, senile systemic (1)	18q11.2-q12.1
Amytrophic lateral sclerosis, one form (2)	21q22.1-q22.2
?Anal canal carcinoma (2)	11q22-qter
Analbuminemia (1)	4q11-q13
?Anemia, megaloblastic, due to DHFR deficiency (1)	5q11.2-q13.2
Anemia, pernicious, congenital, due to deficiency of intrinsic factor (1)	Chr.11
Anemia, sideroblastic/hypochromic (3)	Xp11.21
Aneurysm, familial (1)	2q31

Disorder	*Location*
Angelman syndrome (2)	15q11-q13
Angioedema, hereditary (1)	11q11-q13.1
Anhidrotic ectodermal dysplasia (2)	Xq12.2-13.1
Aniridia of WAGR syndrome (2)	11p13
Aniridia-2 (3)	11p13
?Anophthalmos-1 (2)	Xq27-q28
Anterior segment mesenchymal dysgenesis (2)	4q28-q31
Antithrombin III deficiency (3)	1q23-q25
ApoA-I and apoC-III deficiency, combined (1)	11q23
Apolipoprotein B-100, defective (1)	2p24
[Apolipoprotein H deficiency] (1)	17q23-qter
Argininemia (1)	6q23
Argininosuccinicaciduria (1)	7cen-q11.2
Aspartylglucosaminuria (3)	4q23-q27
Ataxia-telangiectasia (2)	11q22-q23
Atherosclerosis, susceptibility to (2)	19p13.3-p13.2
Atopy (2)	11q12-q13
Atransferrinemia (1)	3q21
Atrial septal defect, secundum type (2)	6p21.3
Autonomic failure due to DBH deficiency (1)	9q34
Basal cell nevus syndrome (2)	9q31
Batten disease (2)	16p12
?Batten disease, one form, 204200 (1)	15q24-q25
Becker muscular dystrophy (3)	Xp21.2
Beckwith-Wiedemann syndrome (2)	11pter-p15.4
Bernard-Soulier syndrome (1)	17pter-p12
11-beta-hydroxysteroid dehydrogenase deficiency (1)	Chr.1
Blepharophimosis, epicanthus inversus and ptosis (2)	3q2
?Bloom syndrome, 210900 (2)	Chr.12
Borjeson-Forssman-Lehmann syndrome (2)	Xq26-q27
Bornholm eye disease (2)	Xq28
Branchiootic syndrome (2)	8q13.3
?Breast cancer (1)	17p13.3
Breast cancer (1)	6q24-q27
Breast cancer, ductal (2)	1p36
Breast cancer, ductal (2)	Chr.13
Breast cancer-1, early onset (2)	17q21
Brody myopathy (1)	Chr.16
Burkitt lymphoma (3)	8q24.12-q24.13
?C1q deficiency (1)	1p36.3-p34.1
?C1q deficiency (1)	1p36.3-p34.1
C1r/C1s deficiency, combined (1)	12p13
C1r/C1s deficiency, combined (1)	12p13
C2 deficiency (3)	6p21.3
C3 deficiency (1)	19p13.3-p13.2
C3b inactivator deficiency (1)	4q25
C4 deficiency (3)	6p21.3
C4 deficiency (3)	6p21.3
C5 deficiency (1)	9q34.1
C6 deficiency (1)	5p13
C7 deficiency (1)	5p13
C8 deficiency, type I (2)	1p32
C8 deficiency, type II (2)	1p32
C9 deficiency (1)	5p13
?Campomelic dysplasia with sex reversal (2)	8q21.4
Carbamoylphosphate synthetase I deficiency (1)	2p
[Carbonic anhydrase I deficiency] (1)	8q22

Table 2—contd

Disorder	*Location*
?Cardiomyopathy (1)	2q35
Cardiomyopathy, dilated, X-linked (3)	Xp21.2
Cat eye syndrome (2)	22q11
?Cataract, congenital total (2)	Xp
Cataract, Coppock-like (3)	2q33-q35
Cataract, Marner type (2)	16q22.1
Cataract, zonular pulverulent (2)	1q2
CD3, zeta chain, deficiency (1)	1p22.1-q21.1
Central core disease of muscle (2)	19q12-q13.2
Centrocytic lymphoma (2)	11q13
Cerebral amyloid angiopathy (1)	20p11
Cerebrotendinous xanthomatosis (2)	2q33-qter
Ceroid lipofuscinosis, neuronal-1, infantile (2)	1p32
Cervical carcinoma (2)	11q13
[CETP deficiency] (1)	16q21
Charcot-Marie-Tooth neuropathy, slow nerve conduction type Ia (2)	17p11.2
Charcot-Marie-Tooth neuropathy, slow nerve conduction type Ib (2)	1q21.2-q23
Charcot-Marie-Tooth neuropathy, X-linked-1, dominant (2)	Xq13
Charcot-Marie-Tooth neuropathy, X-linked-2, recessive (2)	Xp22.2
Cholesteryl ester storage disease (1)	10q24-q25
Chondrodysplasia punctata, X-linked dominant (2)	Xq28
Chondrodysplasia punctata, X-linked recessive (2)	Xp22.3
Choroideremia (2)	Xq21.2
Chronic granulomatous disease, autosomal, due to deficiency of CYBA (3)	16q24
Chronic granulomatous disease due to deficiency of NCF-1 (1)	7q11.23
Chronic granulomatous disease due to deficiency of NCF-2 (1)	1q25
Chronic granulomatous disease, X-linked (3)	Xp21.1
Chronic infections (1)	10q11.2-q21
Citrullinemia (1)	9q34
Cleft palate, X-linked (2)	Xq13-q21.31
?Cleidocranial dysplasia (2)	8q22
CMO II deficiency (1)	8q21
CMO II deficiency (1)	8q21
?Cockayne syndrome (2)	10q21.1
Coffin-Lowry syndrome (2)	Xp22.2-p22.1
Colorblindness, blue monochromatic (3)	Xq28
Colorblindness, deutan (3)	Xq28
Colorblindness, protan (3)	Xq28
Colorblindness, tritan (2)	7q22-qter
Colorectal adenoma (1)	12p12.1
Colorectal cancer (1)	12p12.1
Colorectal cancer, 114500 (3)	17p13.1
Colorectal cancer (1)	18q23.3
Colorectal cancer (1)	5q21
Colorectal cancer (3)	5q21-q22
Combined C6/C7 deficiency (1)	5p13
?Combined variable hypogammaglobulinemia (1)	14q32.33
Congenital absence of vas deferens (1)	7q31.2
Contractural arachnodactyly, congenital (3)	Chr.5
Coproporphyria (1)	Chr.9
?Cornelia de Lange syndrome (2)	3q26.3
{Coronary artery disease, susceptibility to} (1)	6q27
Cortisol resistance (1)	5q31
CR1 deficiency (1)	1q32
Craniosynostosis (2)	7p21.3-p21.2
[Creatine kinase, brain type, ectopic expression of] (2)	14q32
Creutzfeldt-Jakob disease, 123400 (3)	20pter-p12
Crigler-Najjar syndrome (1)	1q21-q23
Crigler-Najjar syndrome, type I, 218800 (1)	Chr.2
?Cryptorchidism (2)	Xp21
?Cutis laxa, marfanoid neonatal type (1)	7q31.1-q31.3
[Cystathioninuria] (1)	Chr.16
Cystic fibrosis (3)	7q31.2
?Cystinuria (2)	14q22
Deafness, conductive, with stapes fixation (2)	Xq13-q21.1
Deafness, low-tone (2)	5q31-q33
Debrisoquine sensitivity (1)	22q11.2-q12.2
Dentinogenesis imperfecta-1 (2)	4q13-q21
Diabetes insipidus, nephrogenic (3)	Xq28
Diabetes insipidus, neurohypophyseal, 125700 (1)	20pter-p12.21
?Diabetes mellitus, insulin-dependent (2)	6p21.3
Diabetes mellitus, insulin-resistant, with acanthosis nigricans (1)	19p13.3-p13.2
Diabetes mellitus, rare form (1)	11p15.5
Diastrophic dysplasia (2)	5q31-q34
DiGeorge syndrome (2)	22q11
Diphenylhydantoin toxicity (1)	1p11-qter
Diphtheria, susceptibility to (1)	5q23
DNA ligase I deficiency (1)	19q13.3
?Dubin-Johnson syndrome (2)	13q34
Duchenne muscular dystrophy (3)	Xp21.2
[Dysalbuminemic hyperthyroxinemia] (1)	4q11-q13
[Dysalbuminemic hyperzincemia] (1)	4q11-q13
Dysfibrinogenemia, alpha types (1)	4q28
Dysfibrinogenemia, beta types (1)	4q28
Dysfibrinogenemia, gamma types (1)	4q28
Dyskeratosis congenita (2)	Xq28
?Dyslexia-1 (2)	15q11
Dysplasminogenemic thrombophilia (1)	6q26-q27
Dysprothrombinemia (1)	11p11-q12
[Dystransthyretinemic hyperthyroxinemia](1)	18q11.2-q12.1
?EEC syndrome (2)	7q11.2-q21.3
Ehlers-Danlos syndrome, type IV (3)	2q31
Ehlers-Danlos syndrome, type VI, 225400 (1)	1p36.3-p36.2
Ehlers-Danlos syndrome, type VIIA1 (3)	17q21.31-q22.05
Ehlers-Danlos syndrome, type VIIA2 (3)	7q21.3-q22.1
?Ehlers-Danlos syndrome, type X (1)	2q34-q36
[Elliptocytosis, Malaysian-Melanesian type] (1)	17q21-q22
Elliptocytosis-1 (3)	1p36.2-p34
Elliptocytosis-2 (2)	1q21
Elliptocytosis-3 (2)	14q22-q23.2
Emery-Dreifuss muscular dystrophy (2)	Xq28
Emphysema (1)	14q32.1
Emphysema due to alpha-2-macroglobulin deficiency (1)	12p13.3-p12.3
Emphysema-cirrhosis (1)	14q32.1
Endocardial fibroelastosis-2 (2)	Xq28
Enolase deficiency (1)	1pter-p36.13
?Eosinophilic myeloproliferative disorder (2)	12p13
Epidermolysis bullosa dystrophica, dominant, 131750 (3)	3p21
Epidermolysis bullosa dystrophica, recessive, 226600 (3)	3p21
Epidermolysis bullosa, Ogna type (2)	8q24
Epidermolysis bullosa simplex, 131900 (3)	17q12-q21
Epidermolysis bullosa simplex, Dowling-Meara type, 131760 (3)	12q11-q13
Epidermolysis bullosa simplex, Dowling-Meara type, 131760 (3)	17q12-q21
Epidermolysis bullosa simplex, generalized, 131900 (1)	12q11-q13
?Epidermolysis bullosa simplex, localized, 131800 (1)	12q11-q13
?Epidermolysis bullosa, Weber Cockayne type, 131800 (2)	12q11-q13
Epidermolytic hyperkeratosis, 113800 (1)	17q21-q22
Epidermolytic hyperkeratosis, 113800 (2)	12q11-q13
Epilepsy, benign neonatal (2)	10q13.2-q13.3
Epilepsy, juvenile myoclonic (2)	6p21.3
Epilepsy, progressive myoclonus (2)	21q22.3
Epithelioma, self-healing, squamous 1, Ferguson-Smith type (2)	9q31
?Erythremia (1)	7q21
Erythremias, alpha- (1)	16pter-p13.3
Erythremias, beta- (1)	11p15.5
Erythroblastosis fetalis (1)	1p36.2-p34
Erythrokeratodermia variabilis (2)	1p36.2-p34
[Euthyroidal hyper- and hypothyroxinemia] (1)	Xq21-q22
Ewing sarcoma (2)	22q12
Exertional myoglobinuria due to deficiency of LDH-A (1)	11p15.4
Fabry disease (3)	Xq22
Facioscapulohumeral muscular dystrophy (2)	4q35
Factor H deficiency (1)	1q32
Factor V deficiency (1)	1q23
Factor VII deficiency (1)	13q34
Factor X deficiency (1)	13q34
Factor XI deficiency (1)	4q35
Factor XII deficiency (1)	5q33-qter
Factor XIIIA deficiency (3)	6p25-p24
Factor XIIIB deficiency (1)	1q31-q32.1
Familial Mediterranean fever (2)	16p13
?Familial Mediterranean fever, 249100 (1)	9q32-q33
?Fanconi anemia (1)	1q41-q42
Fanconi anemia-1 (2)	20q13.2-q13.3
Favism (1)	Xq28
{?Fetal alcohol syndrome} (1)	12q24.2
?Fetal hydantoin syndrome (1)	1p11-qter
?Fibrodysplasia ossificans progressiva (1)	Chr.20
Fish-eye disease (3)	16q22.1
[Fish-odor syndrome] (1)	1q
Fletcher factor deficiency (1)	4q35
Focal dermal hypoplasia (2)	Xp22.31
Friedreich ataxia (2)	9q13-q21.1
Fructose intolerance (1)	9q22
Fucosidosis (1)	1p34
Fumarase deficiency (1)	1q42.1
G6PD deficiency (3)	Xq28
Galactokinase deficiency (1)	17q21-q22
Galactose epimerase deficiency (1)	1p36-p35
Galactosemia (1)	9p13
Galactosialidosis (1)	20q13.1
Gardner syndrome (2)	5q21-q22
Gaucher disease (1)	1q21
Gaucher disease, variant form (1)	10q21-q22

Table 2—contd

Disorder	*Location*
Genitourinary dysplasia (2)	11p13
Gerstmann-Straussler disease, 137440 (3)	20pter-p12
?Gilbert syndrome, 143500 (1)	Chr.2
Glanzmann thrombasthenia, type A (1)	17q21.32
Glanzmann thrombasthenia, type B (1)	17q21.32
Glaucoma, congenital (2)	Chr.11
Glioblastoma multiforme (2)	10p12-q23.2
Glucose/galactose malabsorption (1)	22q11.2-qter
Glutaricaciduria, type II (1)	15q23-q25
Glutaricaciduria, type IIc (1)	Chr.19
Glutathioninuria (1)	22q11.1-q11.2
Glycerol kinase deficiency (2)	Xp21.3-p21.2
Glycogen storage disease VII (1)	1cen-q32
Glycogen storage disease, X-linked hepatic (2)	Xp22.2-p22.1
Glycogen storage disease-3 (3)	1p21
[Glyoxalase II deficiency] (1)	16p13
GM1-gangliosidosis (1)	3p21-p14.2
GM2-gangliosidosis, AB variant (1)	Chr.5
GM2-gangliosidosis, juvenile, adult (1)	15q23-q24
Goeminne TKCR syndrome (2)	Xq28
Goiter, adolescent multinodular (1)	8q24.2-q24.3
?Goldenhar syndrome (2)	7p
Gonadal dysgenesis, XY female type (2)	Xp22-p21
Gonadoblastoma (2)	11p13
?Gonadotropin deficiency (2)	Xp21
Greig craniopolysyndactyly syndrome (3)	7p13
?Growth hormone deficiency, X-linked (2)	Xq21.3-q22
?Gynecomastia, familial, due to increased aromatase activity (1)	15q21.1
Gyrate atrophy of choroid and retina with ornithinemia, B6 responsive or unresponsive (1)	10q26
Harderoporphyrinuria (1)	Chr.9
Hb H mental retardation syndrome (2)	16pter-p13.3
Heinz body anemias, alpha- (1)	16pter-p13.3
Heinz body anemias, beta- (1)	11p15.5
Hemochromatosis (2)	6p21.3
Hemodialysis-related amyloidosis (1)	15q21-q22
Hemolytic anemia due to ADA excess (1)	20q13.11
Hemolytic anemia due to adenylate kinase deficiency (1)	9q34.1
Hemolytic anemia due to bisphosphoglycerate mutase deficiency (1)	7q31-q34
Hemolytic anemia due to G6PD deficiency (1)	Xq28
Hemolytic anemia due to glucosephosphate isomerase deficiency (1)	19cen-q12
Hemolytic anemia due to glutathione peroxidase deficiency (1)	3q11-q12
Hemolytic anemia due to glutathione reductase deficiency (1)	8p21.1
Hemolytic anemia due to hexokinase deficiency (1)	10q22
Hemolytic anemia due to PGK deficiency (1)	Xq13
Hemolytic anemia due to phosphofructokinase deficiency (1)	21q22.3
Hemolytic anemia due to triosephosphate isomerase deficiency (1)	12p13
Hemophilia A (3)	Xq28
Hemophilia B (3)	Xq27.1-q27.2
Hemorrhagic diathesis due to 'antithrombin' Pittsburgh (1)	14q32.1
Hemorrhagic diathesis due to PAI1 deficiency (1)	7q21.3-q22
?Hepatic lipase deficiency (1)	15q21-q23
?Hepatocarcinoma (1)	2q14-q21
Hepatocellular carcinoma (3)	4q32.1
[Hereditary persistence of alpha-fetoprotein] (3)	4q11-q13
?Hereditary persistence of fetal hemoglobin (3)	11p15.5
?Hereditary persistence of fetal hemoglobin, heterocellular, Indian type (2)	7q36
?Hereditary persistence of fetal hemoglobin, Swiss type (2)	Xp11.23
Hers disease, or glycogen storage disease VI (1)	Chr.14
Heterocellular hereditary persistence of fetal hemoglobin (2)	11p15
[Hex A pseudodeficiency] (1)	15q23-q24
?HHH syndrome (2)	13q34
[Histidinemia] (1)	12q22-q23
?Holoprosencephaly (2)	2p21
Holoprosencephaly, type 3 (2)	7q36
?Holt-Oram syndrome (2)	14q23-q24.2
?Holt-Oram syndrome (2)	20p13
Homocystinuria, B6-responsive and nonresponsive types (1)	21q22.3
HPFH, deletion type (1)	11p15.5
HPFH, nondeletion type A (1)	11p15.5
HPFH, nondeletion type G (1)	11p15.5
HPRT-related gout (1)	Xq26-q27.2
?Humoral hypercalcemia of malignancy (1)	12p12.1-p11.2
Huntington disease (2)	4p16.3
Hurler syndrome (1)	4p16.3
Hurler-Scheie syndrome (1)	4p16.3
Hydrocephalus, X-linked (2)	Xq28
Hydrops fetalis, one form (1)	19cen-q12
3-hydroxyacyl-CoA dehydrogenase deficiency (1)	Chr.7
Hyperammonemia due to CTPase deficiency (1)	1p13-p11
Hyperbetalipoproteinemia (1)	2p24
Hypercalcemia, hypocalciuric, familial (2)	3q21-q24
Hypercholesterolemia, familial (3)	19p13.2-p13.1
?Hyperglycinemia, isolated nonketotic, type I (2)	9p22
?Hyperimmunoglobulin G1 syndrome (2)	14q32.33
Hyperkalemic periodic paralysis (3)	17q23.1-q25.3
?Hyperleucinemia-isoleucinemia or hypervalinemia (1)	12pter-q12
Hyperlipoproteinemia I (1)	8p22
Hyperlipoproteinemia, type Ib (1)	19q13.1
Hyperlipoproteinemia, type III (1)	19q13.1
[Hyperphenylalaninemia, mild] (3)	12q24.1
[?Hyperproglucagonemia] (1)	2q36-q37
Hyperproinsulinemia, familial (1)	11p15.5
Hypertriglyceridemia, one form (1)	11q23
Hypertrophic cardiomyopathy, one form, 192600 (3)	14q12
?Hypervalinemia or hyperleucine-isoleucinemia (1)	Chr.19
Hypoalphalipoproteinemia (1)	11q23
Hypobetalipoproteinemia (1)	2p24
[Hypoceruloplasminemia, hereditary] (1)	3q21-q24
Hypofibrinogenemia, gamma types (1)	4q28
Hypogonadism, hypergonadotropic (1)	19q13.32
?Hypogonadotropic hypogonadism due to GNRH deficiency, 227200 (1)	8p21-p11.2
Hypomagnesemia, X-linked primary (2)	Xp22
?Hypomelanosis of Ito (2)	15q11-q13
?Hypomelanosis of Ito (2)	9q33-qter
Hypoparathyroidism, familial (1)	11p15.3-p15.1
Hypoparathyroidism, X-linked (2)	Xq26-q27
?Hypophosphatasia, adult 146300 (1)	1p36.1-p34
Hypophosphatasia, infantile 241500 (3)	1p36.1-p34
Hypophosphatemia, hereditary (2)	Xp22.2-p22.1
?Hypophosphatemia with deafness (2)	Xp22
Hypoprothrombinemia (1)	11p11-q12
?Hypospadias-dysphagia syndrome (2)	5p13-p12
Hypothyroidism, hereditary congenital (1)	8q24.2-q24.3
Hypothyroidism, nongoitrous (1)	1p13
Hypothyroidism, nongoitrous, due to TSH resistance (1)	14q31
?Ichthyosis vulgaris, 146700 (1)	1q21
Ichthyosis, X-linked (3)	Xp22.32
?Immotile cilia syndrome (2)	6p
Immunodeficiency, X-linked, with hyper-IgM (2)	Xq24-q27
Incontinentia pigmenti, familial (2)	Xq27-q28
Incontinentia pigmenti, sporadic type (2)	Xp11.21
Infertile male syndrome (1)	Xcen-q22
[Inosine triphosphatase deficiency] (1)	20p
Insomnia, fatal familial (3)	20pter-p12
Interferon, alpha, deficiency (1)	9p21
Interferon, immune, deficiency (1)	12q24.1
?Isolated growth hormone deficiency due to defect in GHRF (1)	20p11.23-qter
Isolated growth hormone deficiency, Illig type with absent GH and Kowarski type with bioinactive GH (3)	17q22-q24
Isovalericacidemia (1)	15q14-q15
?Jacobsen syndrome (2)	11q
Kallmann syndrome (2)	Xp22.3
[Kappa light chain deficiency] (1)	2p12
Keratosis follicularis spinulosa decalvans (2)	Xp22.2-p21.2
3-ketothiolase deficiency (1)	11q22.3-q23.1
[Kininogen deficiency] (1)	3q26-qter
?Kniest dysplasia (1)	12q13.11-q13.2
?Kostmann agranulocytosis (2)	6p21.3
Krabbe disease (1)	14q21-q31
?Lactase deficiency, adult, 223100 (1)	Chr.2
?Lactase deficiency, congenital (1)	Chr.2
?Lactoferrin-deficient neutrophils, 245480 (1)	3q21-q23
Langer-Giedion syndrome (2)	8q24.11-q24.13
Langer-Saldino achondrogenesis-hypochondrogenesis (1)	12q13.11-q13.2
Laron dwarfism (1)	5p13-p12
?Laryngeal adductor paralysis (2)	6p21.3-p21.2
{Lead poisoning, susceptibility to} (1)	9q34
?Leiomyomata, multiple hereditary cutaneous (2)	18p11.32
Leprechaunism (1)	19p13.3-p13.2
Lesch-Nyhan syndrome (3)	Xq26-q27.2
?Letterer-Siwe disease (2)	13q14-q31
Leukemia, acute lymphoblastic (1)	19p13.3-p13.2
Leukemia, acute lymphoblastic (2)	9p22-p21
?Leukemia, acute lymphocytic, with 4/11 translocation (3)	4q21
Leukemia, acute myeloid (2)	21q22
Leukemia, acute myeloid, M2 type (1)	Xpter-p21
Leukemia, acute pre-B-cell (2)	1q23
Leukemia, acute promyelocytic (1)	17q21.1
Leukemia, acute promyelocytic (2)	15q22
Leukemia, acute T-cell (2)	11p13
Leukemia, chronic myeloid (3)	22q11.21
Leukemia, chronic myeloid (3)	9q34.1
Leukemia, myeloid/lymphoid or mixed-lineage (2)	11q23
Leukemia, T-cell acute lymphoblastic (2)	11p15

Table 2—contd

Disorder	*Location*
Leukemia, T-cell acute lymphoblastic (2)	9q34.3
Leukemia, T-cell acute lymphoblastoid (2)	19p13
Leukemia, T-cell acute lymphocytic (2)	10q24
?Leukemia, transient (2)	21q11.2
Leukemia-1, T-cell acute lymphoblastic (3)	1p32
Leukemia-2, T-cell acute lymphoblastic (3)	9q31
Leukemia/lymphoma, B-cell, 1 (2)	11q13.3
Leukemia/lymphoma, B-cell, 2 (2)	18q21.3
Leukemia/lymphoma, B-cell, 3 (2)	19q13
Leukemia/lymphoma, T-cell (2)	14q32.1
Leukemia/lymphoma, T-cell (2)	2q34
Leukemia/lymphoma, T-cell (3)	14q11.2
Leukocyte adhesion deficiency (1)	21q22.3
Li-Fraumeni syndrome (1)	17p13.1
Lipoamide dehydrogenase deficiency (1)	7q31-q32
Lipoma (2)	12q13-q14
Liver cell carcinoma (1)	11p14-p13
Long QT syndrome (2)	11p15.5
Lowe syndrome (2)	Xq26.1
Lupus erythematosus, systemic, 152700 (1)	1q23
Lymphoproliferative syndrome, X-linked (2)	Xq25
?Lynch cancer family syndrome II (2)	18q11-q12
?Lysosomal acid phosphatase deficiency (1)	11p12-p11
Macrocytic anemia of 5q- syndrome, refractory (2)	5q12-q32
Macular dystrophy, atypical vitelliform (2)	8q24
Macular dystrophy, North Carolina type (2)	6q13-q21
Macular dystrophy, vitelliform type (2)	11q13
?Male infertility due to acrosin deficiency (2)	22q13-qter
?Male infertility, familial (1)	11p13
?Male pseudohermaphroditism due to defective LH (1)	19q13.32
Malignant hyperthermia, 145600 (3)	19q13.1
?Malignant melanoma, cutaneous (2)	1p36
?Manic-depressive illness (2)	11p15.5
?Manic-depressive illness, X-linked (2)	Xq28
Mannosidosis (1)	19p13.2-q12
Maple syrup urine disease, type 1 (3)	19q13.1-q13.2
Maple syrup urine disease, type 2 (3)	1p31
Maple syrup urine disease, type 3 (1)	6p22-p21
Marfan syndrome, 154700 (3)	15q21.1
Maroteaux-Lamy syndrome, several forms (1)	5q11-q13
Martin-Bell syndrome (2)	Xq27.3
MASA syndrome (2)	Xq28
McArdle disease (1)	11q13
McCune-Albright polyostotic fibrous dysplasia, 174800 (1)	20q13.2
[McLeod phenotype] (2)	Xp21.2-p21.1
Medullary thyroid carcinoma (2)	10q21.1
Megacolon (2)	13q22.1-q32.1
Megalocornea, X-linked (2)	Xq21.3-q22
?Melanoma (1)	9p23
Meningioma (2)	22q12.3-qter
Meningioma (3)	22q12.3-q13.1
Menkes disease (2)	Xq12-q13
Mental retardation of WAGR (2)	11p13
Mental retardation, X-linked, syndromic-1, with dystonic movements, ataxia, and seizures (2)	Xp22.2-p22.1
Mental retardation, X-linked, syndromic-2, with dysmorphism and cerebral atrophy (2)	Xp11-q21
Mental retardation, X-linked, syndromic-3, with spastic diplegia (2)	Xp11-q21.3
Mental retardation, X-linked, syndromic-4, with congenital contractures and low fingertip arches (2)	Xq13-q22
Mental retardation, X-linked, syndromic-5, with Dandy-Walker malformation, basal ganglia disease, and seizures (2)	Xq25-q27
Mental retardation, X-linked, syndromic-6, with gynecomastia and obesity (2)	Xp21.1-q22
Mental retardation, X-linked-1, non-dysmorphic (2)	Xp22
Mental retardation, X-linked-2, non-dysmorphic (2)	Xq11-q12
?Mental retardation, X-linked-3 (2)	Xq28
Mental retardation-skeletal dysplasia (2)	Xq28
Metachromatic leukodystrophy (1)	22q13.31-qter
Metachromatic leukodystrophy due to deficiency of SAP-1 (1)	10q21-q22
Methemoglobinemia due to cytochrome b5 deficiency (1)	Chr.18
Methemoglobinemia, enzymopathic (1)	22q13.31-qter
Methemoglobinemias, alpha- (1)	16pter-p13.3
Methemoglobinemias, beta- (1)	11p15.5
Methylmalonicaciduria, mutase deficiency type (1)	6p21
Mevalonicaciduria (1)	Chr.12
Miller-Dieker lissencephaly syndrome (2)	17p13.3
MODY, one form, 125850 (3)	11p15.5
MODY, type I (2)	20q13
MODY, type II, 125851 (3)	7p15-p13
?Moebius syndrome (2)	13q12.2-q13
?Monocyte carboxyesterase deficiency (1)	16q13-q22.1
Mucolipidosis II (1)	4q21-q23
Mucolipidosis III (1)	4q21-q23
Mucopolysaccharidosis I (1)	4p16.3
Mucopolysaccharidosis II (2)	Xq28
Mucopolysaccharidosis IVB (1)	3p21-p14.2
Mucopolysaccharidosis VII (1)	7q21.11
Multiple endocrine neoplasia I (1)	11q13
Multiple endocrine neoplasia II (2)	10q21.1
Multiple endocrine neoplasia III(2)	10q21.1
?Multiple exostoses (2)	8q23-q24.1
?Multiple lipomatosis (2)	12q13-q14
?Muscle glycogenosis (1)	Xq12-q13
Muscular dystrophy, limb-girdle, autosomal dominant (2)	5q22.3-q31.3
Muscular dystrophy, limb-girdle, autosomal recessive (2)	15q15-q22
Myeloperoxidase deficiency (1)	17q21.3-q22
Myoadenylate deaminase deficiency (1)	1p21-p13
Myoglobinuria/hemolysis due to PGK deficiency (1)	Xq13
Myopathy due to CTPase deficiency (1)	1p13-p11
Myopathy due to phosphoglycerate mutase deficiency (1)	7p13-p12.3
Myopia-1 (2)	Xq28
Myotonia congenita, atypical acetazolamide-responsive (3)	17q23.1-q25.3
Myotonic dystrophy (2)	19q13.2-q13.3
Myotubular myopathy, X-linked (2)	Xq28
Myxoid liposarcoma (2)	12q13-q14
?N syndrome, 310465 (1)	Xp22.3-p21.1
Nail-patella syndrome (2)	9q34
Nance-Horan syndrome (2)	Xp22.3-p21.1
Nemaline myopathy (2)	1q21-q23
Neuroblastoma (2)	1p36.2-p36.1
Neuroepithelioma (2)	22q12
Neurofibromatosis, von Recklinghausen (2)	17q11.2
Neutropenia, immune (2)	1q23
Niemann-Pick disease (1)	11p15.4-15.1
Nightblindness, congenital stationary, type I (2)	Xp11.3
Norrie disease (2)	Xp11.4
Norum disease (3)	16q22.1
Nucleoside phosphorylase deficiency, immunodeficiency due to (1)	14q13.1
?Obesity (2)	7q31
?Ocular albinism autosomal recessive (2)	6q13-q15
Ocular albinism, Forsius-Eriksson type (2)	Xp11-q11
Ocular albinism, Nettleship-Falls type (2)	Xp22.3
?Oculocutaneous albinism, type II (2)	2q31.2
Ornithine transcarbamylase deficiency (3)	Xp21.1
Orofacial cleft (2)	6pter-p23
Oroticaciduria (1)	3q13
Osteoarthrosis, precocious (3)	12q13.11-q13.2
Osteogenesis imperfecta, 2 or more clinical forms (3)	17q21.31-q22.05
Osteogenesis imperfecta, 2 or more clinical forms (3)	7q21.3-q22.1
?Osteopetrosis, 259700 (1)	1p21-p13
Osteosarcoma, retinoblastoma-related (2)	13q14.1
Otopalatodigital syndrome, type I (2)	Xq28
Ovarian carcinoma (2)	9p24
Ovarian failure, premature (2)	Xq26-q27
Oxalosis I (1)	2q36-q37
?Paget disease of bone (2)	6p21.3
Pancreatic lipase deficiency (1)	10q26.1
?Panhypopituitarism (1)	3q
Paraganglioma (2)	11q23-qter
Paramyotonia congenita (3)	17q23.1-q25.3
Parathyroid adenomatosis 1 (2)	11q13
{?Parkinsonism, susceptibility to} (1)	22q11.2-q12.2
Paroxysmal nocturnal hemoglobinuria (1)	11p14-p13
Pelizaeus-Merzbacher disease (3)	Xq22
?Pendred syndrome (2)	8q24
Periodontitis, juvenile (2)	4q11-q13
Persistent Mullerian duct syndrome (1)	19p13.3-p13.2
Phenylketonuria (3)	12q24.1
Phenylketonuria due to dihydropteridine reductase deficiency (1)	4p15.31
Phosphoribosylpyrophosphate synthetase-related gout (1)	Xq22-q24
?Phosphorylase kinase deficiency of liver and muscle, 261750 (2)	16q12-q13.1
Piebaldism (3)	4q11-q12
Pituitary tumor, growth-hormone-secreting (1)	20q13.2
PK deficiency hemolytic anemia (1)	1q21
[Placental lactogen deficiency] (1)	17q22-q24
Placental steroid sulfatase deficiency (3)	Xp22.32
Plasmin inhibitor deficiency (1)	17pter-p12
Plasminogen activator deficiency (1)	8p12
Plasminogen deficiency, types I and II (1)	6q26-q27
Plasminogen Tochigi disease (1)	6q26-q27
Platelet alpha/delta storage pool deficiency (1)	1q21-q24
{Polio, susceptibility to} (2)	19q12-q13.2
Polycystic kidney disease (2)	16p13.31-p13.12
Polycystic ovarian disease (1)	17q11-q12
Polyposis coli, familial (2)	5q21-q22
Pompe disease (1)	17q23
Porphyria, acute hepatic (1)	9q34

Table 2—contd

Disorder	*Location*
Porphyria, acute intermittent (1)	11q24.1-q24.2
Porphyria, Chester type (2)	11q
Porphyria, congenital erythropoietic (1)	10q25.2-q26.3
Porphyria cutanea tarda (1)	1p34
Porphyria, hepatoerythropoietic (1)	1p34
Porphyria variegata (2)	14q32
Postanesthetic apnea (1)	3q26.1-q26.2
Prader-Willi syndrome (2)	15q11
Progressive cone dystrophy (2)	Xp21.1-p11.3
Prolidase deficiency (1)	19cen-q13.11
Properdin deficiency, X-linked (3)	Xp11.4-p11.23
Propionicacidemia, type I or pccA type (1)	13q32
Propionicacidemia, type II or pccB type (1)	3q21-q22
Protein C deficiency (1)	2q13-q14
Protein S deficiency (1)	3p11.1-q11.2
Protoporphyria, erythropoietic (1)	18q21.3
Pseudohermaphroditism, male, with gynecomastia (1)	17q11-q12
Pseudohypoaldosteronism (1)	4q31.1
Pseudohypoparathyroidism, type Ia (1)	20q13.2
Pseudo-vitamin D dependency rickets 1 (2)	12q14
Pseudo-Zellweger syndrome (1)	3p23-p22
?Pyridoxine dependency with seizures (1)	2q31
Pyropoikilocytosis (1)	1q21
Pyruvate carboxylase deficiency (1)	11q
Pyruvate dehydrogenase deficiency (1)	Xp22.2-p22.1
?Rabson-Mendenhall syndrome (1)	19p13.3-p13.2
?Ragweed sensitivity (2)	6p21.3
Reifenstein syndrome (1)	Xcen-q22
Renal cell carcinoma (2)	3p14.2
[Renal glucosuria] (2)	6p21.3
Renal tubular acidosis-osteopetrosis syndrome (1)	8q22
?Retinal cone dystrophy-1 (2)	6q25-q26
?Retinal cone-rod dystrophy (2)	18q21-q22.2
Retinitis pigmentosa, autosomal dominant (3)	3q21-q24
Retinitis pigmentosa, peripherin-related (3)	6p21.1-cen
Retinitis pigmentosa-1 (2)	8p11-q21
Retinitis pigmentosa-2 (2)	Xp11.3
Retinitis pigmentosa-3 (2)	Xp21.3-p21.1
Retinoblastoma (2)	13q14.1-q14.2
?Retinol binding protein, deficiency of (1)	10q23-q24
Retinoschisis (2)	Xp22.3-p22.1
?Rett syndrome (2)	Xp
Rhabdomyosarcoma (2)	11p15.5
Rhabdomyosarcoma, alveolar (2)	2q37
Rh-null disease (1)	3cen-q22
?Rh-null hemolytic anemia (1)	1p36.2-p34
Rickets, vitamin D-resistant (1)	12q12-q14
Rieger syndrome (2)	4q23-q27
Rod monochromacy (2)	Chr.14
?Rothmund-Thomson syndrome (2)	Chr.8
Rubinstein-Taybi syndrome (2)	16p13.3
Salivary gland pleomorphic adenoma (2)	8q12
Sandhoff disease (1)	5q13
?Sanfilippo disease, type IIIC (2)	Chr.14
Sanfilippo syndrome D (1)	12q14
Sarcoma, synovial (2)	Xp11.2
Scheie syndrome (1)	4p16.3
?Schizophrenia (2)	5q11.2-q13.3
Sclerotylosis (2)	4q28-q31
Severe combined immunodeficiency due to ADA deficiency (1)	20q13.11
Severe combined immunodeficiency due to IL2 deficiency (1)	4q26-q27
Severe combined immunodeficiency, HLA class II-negative type (1)	19p13.1
Severe combined immunodeficiency, X-linked (2)	Xq13.1-q21.1
?Sialidosis (2)	6p21.3
Sickle cell anemia (1)	11p15.5
?Simpson-Galabi-Behmel syndrome (2)	Xcen-q21.3
?Situs inversus viscerum (2)	14q32
?SLE (1)	1q32
Small-cell cancer of lung (2)	3p23-p21
?Smith-Lemli-Opitz syndrome (2)	7q34-qter
Smith-Magenis syndrome (2)	17p11.2
Spastic paraplegia, X-linked, uncomplicated (2)	Xq21-q22
Spherocytosis, hereditary, Japanese type (1)	15q15
Spherocytosis, recessive (1)	1q21
Spherocytosis-1 (3)	14q22-q23.2
Spherocytosis-2 (3)	8p11.2
Spinal and bulbar muscular atrophy of Kennedy, 313200 (3)	Xcen-q22
Spinal muscular atrophy II (2)	5q12.2-q13.3
Spinal muscular atrophy III (2)	5q12.2-q13.3
Spinocerebellar ataxia-1 (2)	6p21.3-p21.2
Split-hand/foot deformity, type 1 (2)	7q21.2-q21.3
Spondyloepiphyseal dysplasia congenita (3)	12q13.11-q13.2
Spondyloepiphyseal dysplasia tarda (2)	Xp22
Startle disease (2)	5q33-q35
Stickler syndrome (3)	12q13.11-q13.2
Sucrose intolerance (1)	3q25-q26
?Susceptibility to amyloid in FMF, 249100 (1)	11pter-p12
Tay-Sachs disease (1)	15q23-q24
Testicular feminization (1)	Xcen-q22
Thalassemias, alpha- (1)	16pter-p13.3
Thalassemias, beta- (1)	11p15.5
Thrombocytopenia, X-linked (2)	Xp21-p11
Thrombophilia due to elevated HRG (2)	3p14-qter
Thrombophilia due to excessive plasminogen activator inhibitor (1)	7q21.3-q22
Thrombophilia due to heparin cofactor II deficiency (1)	22q11
Thyroid hormone resistance, 274300, 188570 (3)	3p24.3
Thyroid iodine peroxidase deficiency (1)	2pter-p12
Thyroid papillary carcinoma (1)	10q11-q12
Thyrotropin-releasing hormone deficiency (1)	Chr.3
Torsion dystonia (2)	9q32-q34
Torsion dystonia-parkinsonism, Filipino type (2)	Xq12-q21.1
?Tourette syndrome (2)	18q22.1
Transcobalamin II deficiency (1)	22q11.2-qter
[Transcortin deficiency] (1)	14q31-q32.1
Treacher Collins mandibulofacial dysostosis (2)	5q31.3-q33.3
Trichorhinophalangeal syndrome, type I (2)	8q24.12
Trypsinogen deficiency (1)	7q32-qter
Tuberous sclerosis-1 (2)	9q33-q34
?Tuberous sclerosis-2 (2)	11q23
?Tuberous sclerosis-3 (2)	12q23.3
Tuberous sclerosis-4 (2)	16p13
Turner syndrome (1)	Xq13.1
Tyrosinemia, type I (1)	15q23-q25
Tyrosinemia, type II (1)	16q22.1-q22.3
Urolithiasis, 2,8-dihydroxyadenine (1)	16q24
Usher syndrome, type 1A (2)	14q
Usher syndrome, type 2 (2)	1q32
van der Woude syndrome (2)	1q32
?Velocardiofacial syndrome (2)	22q11
Vitreoretinopathy, exudative, familial (2)	11q13-q23
{Vivax malaria, susceptibility to} (1)	1q21-q22
von Hippel-Lindau syndrome (2)	3p26-p25
von Willebrand disease (1)	12pter-p12
Waardenburg syndrome, type I (3)	2q35
Waisman parkinsonism-mental retardation syndrome (2)	Xq28
Watson syndrome (2)	17q11.2
Werdnig-Hoffmann disease (2)	5q12.2-q13.3
Werner syndrome (2)	8p12-p11
Wieacker-Wolff syndrome (2)	Xq13-q21
Wilms tumor (2)	11p13
Wilms tumor, type 2 (2)	11p15.5
Wilson disease (2)	13q14-q21
Wiskott-Aldrich syndrome (2)	Xp11.3-p11.2
?Wolf-Hirschhorn syndrome (3)	4p16.1
Wolman disease (1)	10q24-q25
?Xeroderma pigmentosum (1)	1q41-q42
Xeroderma pigmentosum, group B (1)	2q21
Xeroderma pigmentosum, group D, 278730 (1)	19q13.2-q13.3
?Xeroderma pigmentosum, one type (1)	Chr.13
Xeroderma pigmentosum, type A (1)	9q34.1
Xeroderma pigmentosum, type F (2)	Chr.15
46,XY female (2)	Xp22.2-p21.3
Zellweger syndrome-1 (2)	7q11.23