Review article

The morbid anatomy of the human genome: chromosomal location of mutations causing disease (update 1 December 1993)

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A survey of the chromosomal location of mutations causing genetic disorders was published in this journal in January 1993 (McKusick and Amberger, 1993). The information presented then was complete to 1 November 1992. Here, we present information that has become available since that date, including both new assignments and revision of previous assignments. This information on the so-called morbid anatomy of the human genome is derived from a synopsis of the human gene map that has been updated continuously since 1971 as part of McKusick's *Mendelian inheritance in man* (10th ed, 1992) and of OMIM (online *Mendelian inheritance in man*, available generally since 1987).

In table 1, new 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. The new information is presented in pictorial form in the figure.

In table 2 an alphabetised list of these disorders and the chromosomal location of the mutation in each case are provided. After the name of the disorder, both in the "Disorder" field of table 1 and in table 2, the numbers 1, 2, or 3 in parentheses indicate that its chromosomal location was determined by mapping of the wild type gene (1), by mapping of the clinical phenotype (2), or by both strategies (3). (In some instances, a (3) is used when the clinical phenotype has been mapped and an intragenic mutation has been identified in a candidate gene even though the wild type gene has not been independently mapped.)

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 included in the figure.

Title – name of gene locus

MIM# – This is the number in McKusick's *Mendelian inheritance in man* (MIM, 10th ed, 1992), its continuously updated online version OMIM, and its periodically released CD-ROM version. (For historical reasons, the number may sometimes indicate location of the entry in the "dominant catalogue" because the wild type gene was characterised and mapped before the recessive disorder resulting from mutation at that site. The practice has been to create only one entry in MIM for each gene locus; however, for some disorders, an entry describing the phenotype, identified by a number sign (#), and a separate entry, identified by an asterisk, for the protein affected by the mutation have been created. Marfan syndrome (154700) and fibrillin (134797) are examples; see chromosome 15.)

Method of mapping

A = in situ DNA-RNA or DNA-DNA annealing ("hybridisation"); for example, COL7A1 to 3p21.3 2.

C = chromosome mediated gene transfer (CMGT); for example, cotransfer of COL1A1 and thymidine kinase gene on chromosome 17.

Ch = chromosomal change associated with particular phenotype and not proved to represent linkage (Fc), deletion (D), or virus effect (V); for example, blepharophimosis, epicanthus inversus, and ptosis to 3q22–q23.

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.

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

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linked loci are identified by a DNA polymorphism, Fd is used; for example, Osler-Rendu-Weber disease on chromosome 9q. F=L in the symbolism of the HGM workshops.

H = based on presumed homology; for example, Neimann-Pick disease, type C, on 19p. Mainly heuristic or confirmatory.

LD = linkage disequilibrium; includes homozygosity mapping, for example, familial dysautonomia.

M = Microcell mediated gene transfer (MMGT); for example, Bloom syndrome.

Psh = PCR of somatic cell hybrid DNA.

RE = Restriction endonuclease techniques; for example, physical linkage of 3 fibrinogen genes (on 4q) and apolipoprotein cluster, including APOC3 on 11q.

REa = combined with somatic cell hybridisation; for example, gene for type VII collagen (COL7A1), which is mutant in dystrophic epidermolysis bullosa.

REb = combined with chromosome sorting; for example, thyroglobulin to 8q. (For this method, using flow sorted chromosomes, W was the symbol adopted by the HGM workshops.)

REn = neighbour analysis in restriction fragments, for example, in pulsed-field gel electrophoresis (PFGE); for example, protein C inhibitor.

S = 'segregation' (cosegregation) of human cellular traits and human chromosomes (or segments of chromosomes) in particular clones from interspecies somatic cell hybrids; for example, GM2 ganglioside activator protein on chromosome 5. When with restriction enzyme, REa.

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) = wild type gene mapped. (2) = disease phenotype mapped. (3) = both wild type gene and disease phenotype mapped (or disease phenotype mapped and mutations identified in the wild type gene).

Mouse – Mouse chromosome carrying homologous gene.

Discussion

With the additions made in the last year, a total of 928 disorders have by now been 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 in this count.) The 928 disorders are distributed over 767 loci; many loci have more than one allelic mutation producing distinct phenotypes.

Recent examples of disorders that have been mapped to specific sites by virtue of mapping of the wild type gene (labelled with the number 1) include Crigler-Najjar syndrome, glycogen storage disease, type I, and metaphyseal chondrodysplasia, Schmid type.

Recent examples of disorders (labelled with the number 2) that have been mapped only on the basis of the clinical phenotype, which is found to be linked to markers at a particular chromosomal site or is found in association with a chromosomal aberration, include alkaptonuria, cartilage-hair hypoplasia, familial dysautonomia, hereditary haemorrhagic telangiectasia, and Werner syndrome. Rubinstein-Taybi syndrome was mapped on the basis of a chromosomal aberration. The mapping information provides the starting point for isolating the mutant gene by positional cloning or the candidate gene approach.

In the case of yet other disorders (labelled with the number 3) the clinical disorder and the wild type gene have been separately mapped. These include instances in which the candidate gene approach has led to definition of the basic defect in the clinical disorder. Examples in the last year included familial amyotrophic lateral sclerosis, type I (wild type gene = SOD1), supravalvar aortic stenosis (wild type gene = elastin), stenosis of the aqueduct of Sylvius (wild type gene = L1CAM), X linked Charcot-Marie-Tooth disease (wild type gene = connexin-32), and X linked immunodeficiency with hyper-IgM (wild type gene = CD40 ligand).

Mapping of genetic disorders has shed light on both allelism and non-allelism. Examples of non-allelic genetic heterogeneity found or extended in the last year include the following: the number of distinguished types of Charcot-Marie-Tooth disease were increased by the mapping of forms to 1p and 8q; in addition to the forms already mapped, two types of familial hypertrophic cardiomyopathy were mapped to chromosomes 15 and 11; a third form of spinocerebellar ataxia was located to chromosome 14; non-allelic heterogeneity in multiple exostoses and Wagner syndrome was established by finding linkage in some families to chromosome 8 or 12, respectively, but not in others. The practice is to assign a number to the various types of a particular disorder as they are defined; for example, CMH1-4 for the forms of hypertrophic cardiomyopathy. The number of forms of retinitis pigmentosa defined on the basis of linkage has now reached RP10. As soon as the nature of the gene that is mutated is identified, one can substitute for the number a designation such as "retinitis pigmentosa, rhodopsin-related" or "retinitis pigmentosa, peripherin-related."

Allelic series as the basis of phenotypic diversity, that is, allelic heterogeneity, is illustrated by pairs or sets of phenotypes such as cystic fibrosis and congenital bilateral absence of vas deferens (caused by mutation in the CFTR gene) and the some seven disorders that have been related to mutations in the type II collagen gene. For the most part, refined delineation of allelic heterogeneity has come not from mapping but rather through detection of point mutations and genotype-phenotype correlations.

Some sets of disorders show both non-allelic genetic heterogeneity and allelic genetic heterogeneity in a symmetrical relationship. The earliest and best known example comes from

the type I collagen genes: certain forms of osteogenesis imperfecta and of Ehlers-Danlos syndrome result from mutation in either the COL1A1 gene on chromosome 17 or the COL1A2 gene on chromosome 7. A more recently elucidated example is epidermolysis bullosa simplex (EBS) which in any one of the distinct Koebner, Dowling-Meara, or Weber-Cockayne forms can be caused by mutation in either the keratin-5 gene on chromosome 12 or the keratin-14 gene on chromosome 17. The heterotrimeric structure of type I collagen constituted by polypeptide chains encoded by the COL1A1 and COL1A2 genes and the close structural-functional association of the K5 and K14 genes in the epidermis are the basis.

Mapping information suggest 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. Tissue-specific alternative splicing of RNA might account for different patterns of involved organs depending on the site of the mutation in the gene. 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. In the last year, Williams syndrome was identified as a probable contiguous gene syndrome, with the elastin gene as one of the genes affected.

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. Recent examples include acute myeloid leukaemia resulting from mutation in the pseudoautosomal CSF2RA gene and paroxysmal nocturnal haemoglobinuria resulting from mutation in the X linked PIGA gene. Some congenital malformations also represent somatic cell genetic disorders. The Happle hypothesis holds that some disorders are the result of 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 the result of 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 11 Human Gene Mapping Workshops 1973 to 1991 and in the recent Human Genome Mapping Workshop 1993 in Kobe, Japan, as well as to others who have assisted in the collation of data on the human gene map, and, of course, to all who have been responsible for generating data on the mapping of genetic disorders. The development and maintenance of *Mendelian inheritance in man* and its online version OMIM were supported in part by the Howard Hughes Medical Institute from 1985 to 1991. It is now supported jointly by the National Center for Human Genome Research of NIH and by the DOE as part of the Genome Data Base (GDB), the repository for mapping information coming from the Human Genome Project.

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

References

1 McKusick VA *Mendelian inheritance in man.* 10th ed. Baltimore: The Johns Hopkins University Press, 1992. (This print version of MIM is published each two years. The 11th edition will be available in mid-1994. *Mendelian inheritance in man* in updated form is also released each 6 months on CD-ROM by Johns Hopkins University Press and Aries Systems Corporation.)

2 References for the new mapping information and additional information about the loci and disorders are provided in the continuously updated OMIM™ (Online *Mendelian inheritance in man*). To obtain information on accessing OMIM, contact:

In the United States: GDB/OMIM User Support, Genome Data Base, Johns Hopkins University, 2024 E. Monument Street, Baltimore, MD 21205-2100, USA (Telephone: 410-955-7058, FAX: 410-614-0434, Internet: help@gdb.org).

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, Internet: cbates@ uk.ac. crc).

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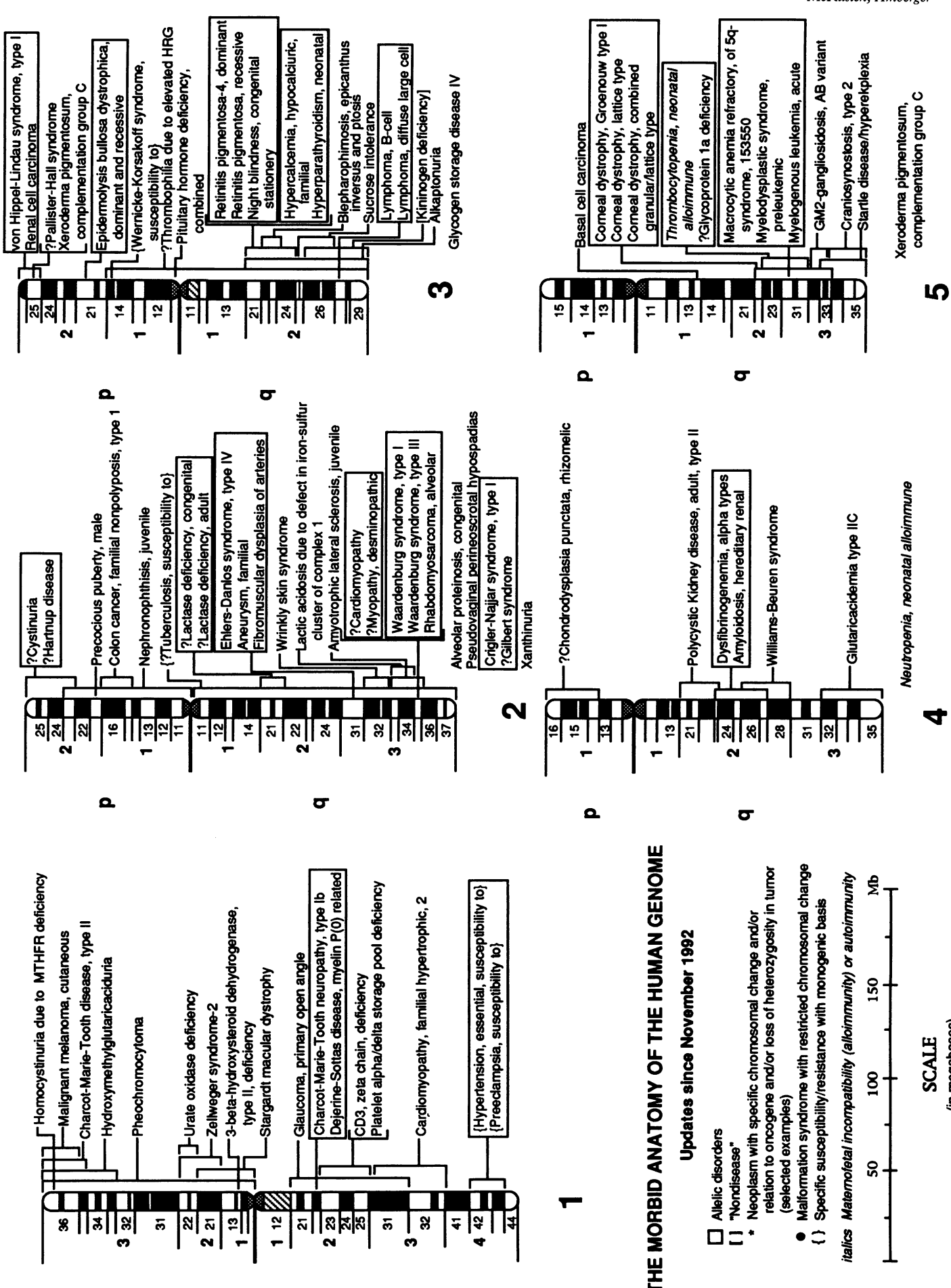
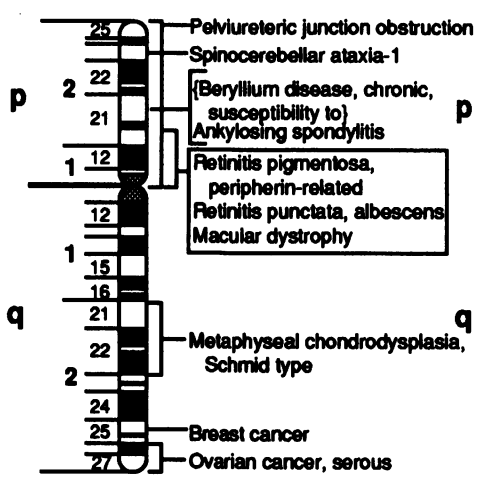
In The Netherlands: GDB User Support, CAOS/CAMM Center, Faculty of Science, University of Nijmegen, PO Box 9010, 6500 GL Nijmegen, Netherlands (Telephone: 31-80-653-391, FAX: 31-652-977, Internet: post@caos.caos.kun.nl).

In Sweden: GDB User Support, Biomedical Center, Box 570, S-751 23 Uppsala, Sweden (Telephone: 46-18-174-057, FAX: 46-18-524-869, Internet: help@gdb.embnet.se).



The morbid anatomy of the human genome: New information acquired in the last year is presented. 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.



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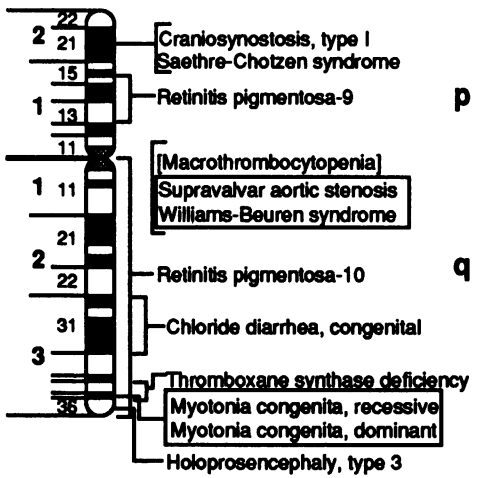

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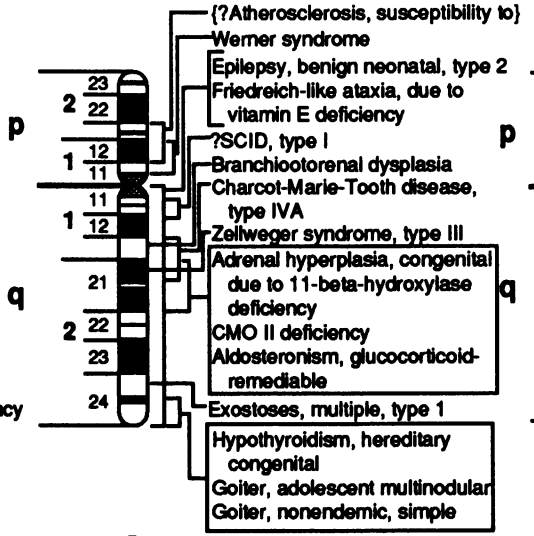

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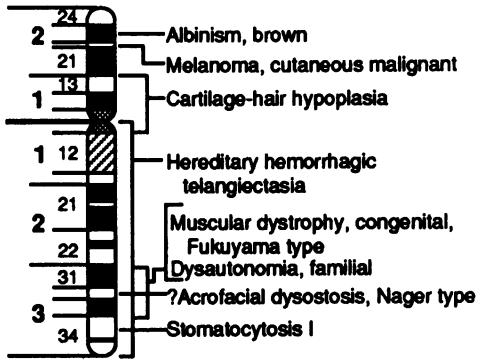

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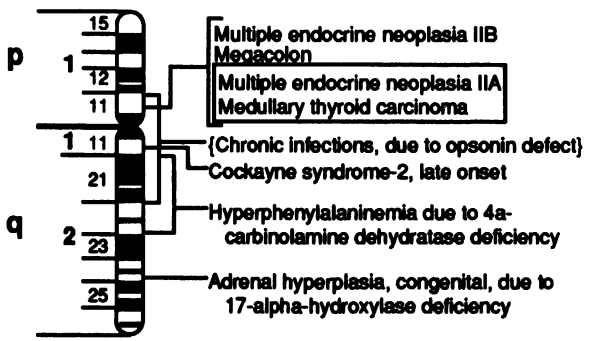

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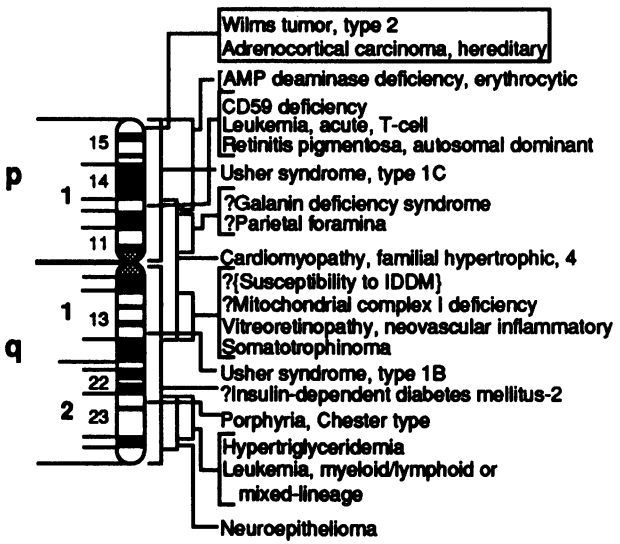

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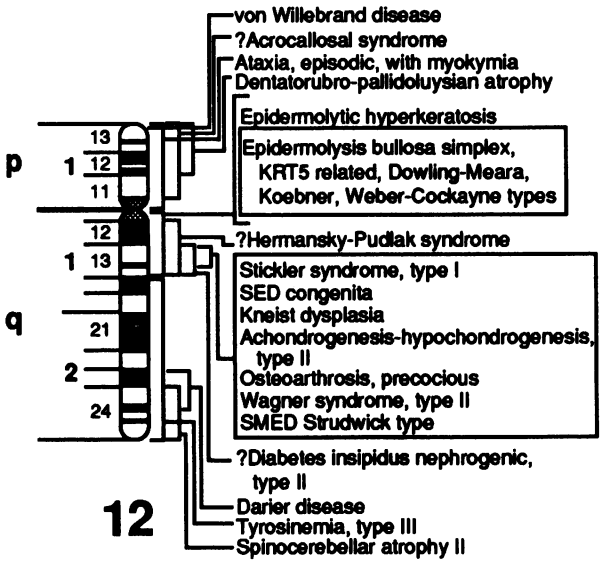

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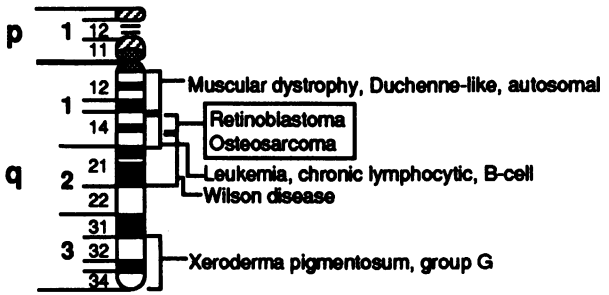
Amyloidosis, renal



Carboxypeptidase B deficiency

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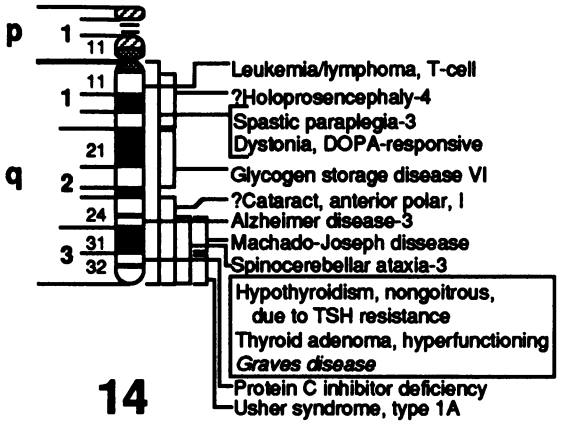

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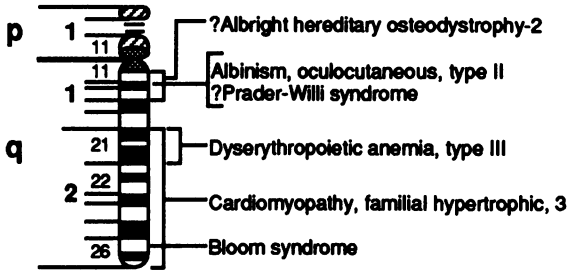

?Xeroderma pigmentosum, type F

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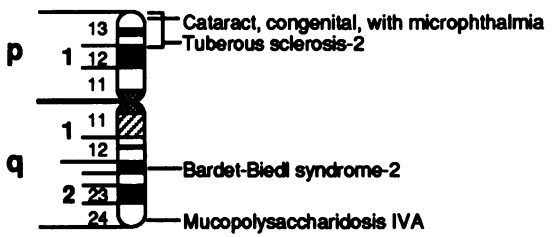

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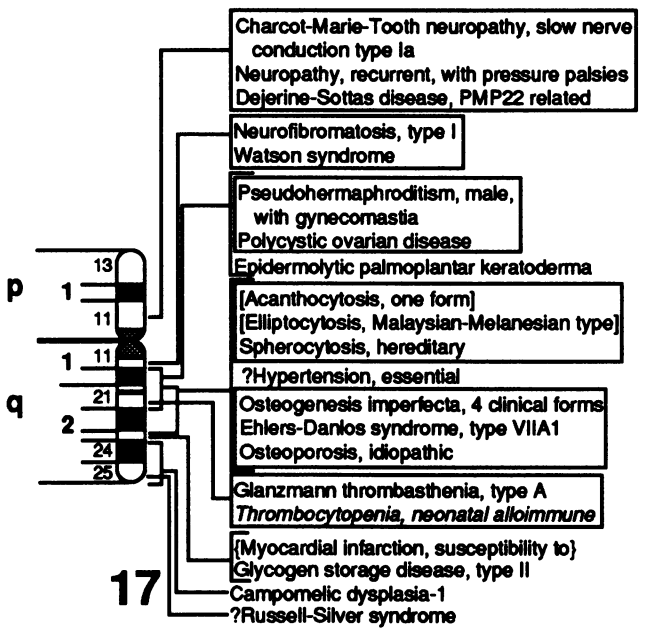

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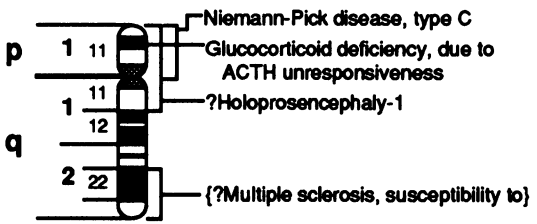

Familial expansile osteolysis

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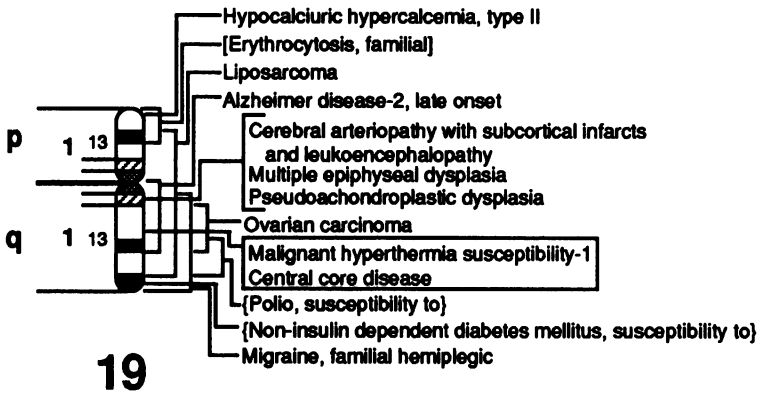
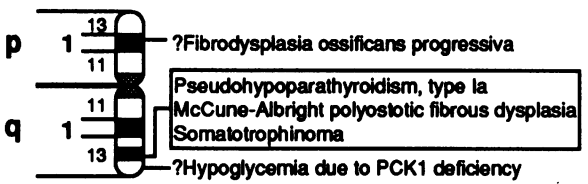
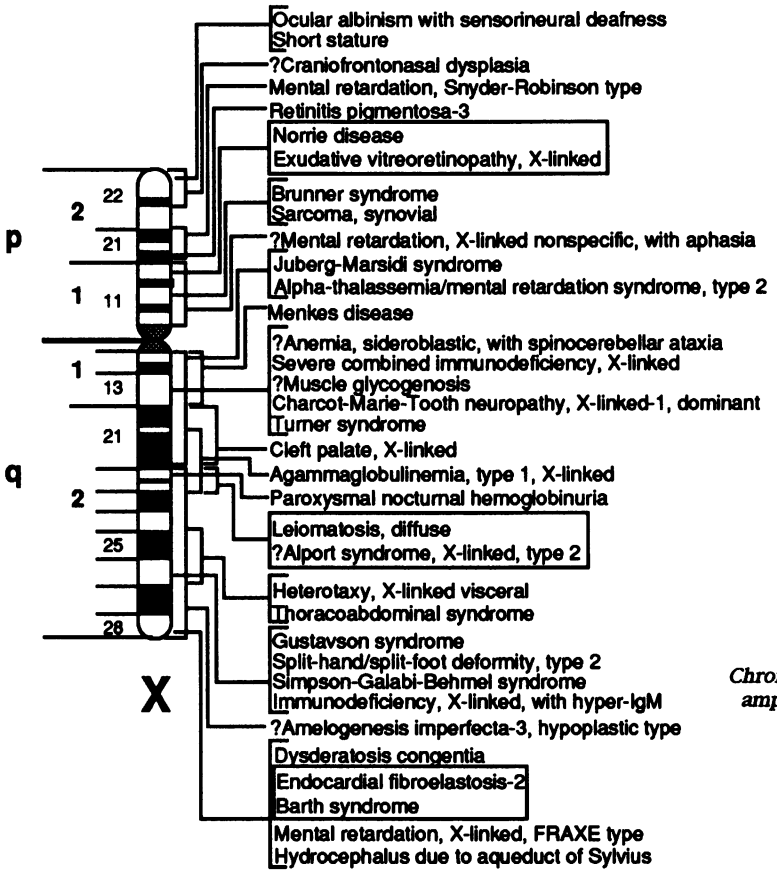
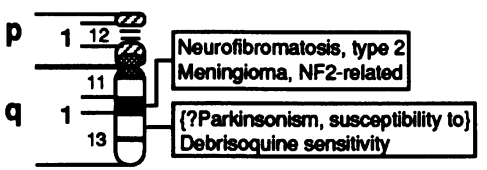


Glutaricaciduria, type IIB





Chromosome amplified 25%



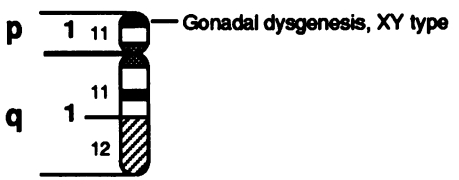


Table 1 The morbid anatomy of the human genome (by chromosome)

Location	Symbol	Status	Title	MIM #	Method	Disorder	Mouse
1pter-p33	HMGCL	P	3-hydroxy-3-methylglutaryl CoA lyase	246450	REa, A	Hydroxymethylglutaricaciduria (1)	
1p36.3	MTHFR	P	Methylenetetrahydrofolate reductase	236250	A	Homocystinuria due to MTHFR deficiency (3)	
1p36	CMM, HCMM, MLM, DNS	P	Cutaneous malignant melanoma/ dysplastic nevus	155600	F, Fd, D	Malignant melanoma, cutaneous (2)	
1p36-p35	CMT2	P	Charcot-Marie-Tooth neuropathy-2 (hereditary motor sensory neuropathy II)	118210	Fd	Charcot-Marie-Tooth disease, type II (2)	
1p22	UOX	P	Urate oxidase	191540	REa, A	Urate oxidase deficiency (1)	
1p22-p21	PXMP1, PMP70	P	Peroxisomal membrane protein-1 (70kD)	170995	REa, A	Zellweger syndrome-2 (1)	3(Pmp70)
1p13.1	HSD3B2	C	Hydroxy-delta-5-steroid steroid delta-isomerase, type 2 (adrenal, gonadal)	201810	A	3-beta-hydroxysteroid deficiency (3) dehydrogenase, type II	3(Hsd3b2)
1p	PCHC	P	Pheochromocytoma	171300	D	Pheochromocytoma (2)	
1p21-p13	STGD	P	Stargardt macular dystrophy	248200	Fd	Stargardt macular dystrophy (2)	
1q21-q31	POAG, GPOA	P	Glaucoma, primary open angle	137760	Fd	Glaucoma, primary open angle, juvenile-onset (2)	
1q22	MPZ, MPP, CMT1B	C	Myelin protein zero	159440	REb, A, F, Fd, D	Charcot-Marie-Tooth neuropathy, 118200 (3); , slow nerve conduction type Ib, Dejerine-Sottas disease, myelin P(0) -related, 145900 (3)	1(Mpp)
1q23-q21.1	CD3Z, TCRZ	C	Antigen CD3Z, zeta polypeptide (TiT3 complex)	186780	REa, A, REn	CD3, zeta chain, deficiency (1)	1(T3z, Cd3z)
1q23-q25	SELP, GRMP	C	Selectin P (granulocyte membrane protein, 140kD; antigen CD62)	173610	REn, A	Platelet alpha/delta storage pool deficiency (1)	1(Grmp)
1q3	CMH2	P	Cardiomyopathy, familial hypertrophic, 2	115195	Fd	Cardiomyopathy, familial hypertrophic, 2 (2)	
1q42-q43	AGT	C	Angiotensinogen	106150	A, REa	{Hypertension, essential, susceptibility to} (3); {Preeclampsia, susceptibility to} (3)	8(Agt)
2pter-q32.3	ATR1, D2H	P	Amino acid-transporter-1	104614	REa	?Cystinuria, 220100 (1); ?Hartnup disease, 234500 (1)	
2p23-cen	NPH1	P	Nephronophthisis-1 (juvenile)	256100	Fd	Nephronophthisis, juvenile (2)	
2p21	LHCGR	P	Luteinizing hormone/ choriogonadotropin receptor	152790	A	Precocious puberty, male, 176410 (1)	
2p16-p15	COCA1, FCC1, MSH	P	Colon cancer, familial, nonpolyposis type 1	120435	Fd	Colon cancer, familial, nonpolyposis type 1 (3)	
2q	TBS, BCG	L	Mycobacterial infections, susceptibility to	209950	H, Fd	{?Tuberculosis, susceptibility to} (2)	
2q21	LCT, LAC, LPH	C	Lactase	223000	REa, Fd, A, Psh	?Lactase deficiency, congenital (1); ?Lactase deficiency, adult, 223100 (1)	
2q31	COL3A1	C	Collagen, type III, alpha-1 polypeptide	120180	REa, A	Ehlers-Danlos syndrome, type IV, 130050 (3); Aneurysm, familial, 100070 (1); Fibromuscular dysplasia of arteries, 135580 (1)	1(Col3a-1)
2q32	WSS	P	Wrinkly skin syndrome	278250	Ch	Wrinkly skin syndrome (2)	
2q33-q34	NDUFS1	P	NADH dehydrogenase ubiquinone), Fe-S protein-1 (75kD)	157655	A	Lactic acidosis due to defect in iron-sulfur cluster of complex I (1)	
2q33-q35	ALS2, ALSJ	P	Amyotrophic lateral sclerosis-2 (juvenile)	205100	Fd	Amyotrophic lateral sclerosis, juvenile (2)	
2q35	DES	P	Desmin	125660	REa, A	?Cardiomyopathy (1); ?Myopathy, desminopathic (1)	1(Des)
2q35	PAX3, WS1, HUP2	C	Paired box homeotic gene-3	193500	Ch, Fd, H, A	Waardenburg syndrome, type I (3); Waardenburg syndrome, type III, 148820 (3); Rhabdomyosarcoma, 148820 (3); Rhabdomyosarcoma, 1(Sp)	
Chr.2	SFTP3	C	Pulmonary surfactant-associated protein-3, 18kD	178640	REa	Alveolar proteinosis, congenital, 265120 (1)	6(Sftp-3)
Chr.2	SRD5A2	P	Steroid-5-alpha-reductase, alpha polypeptide-2 (3-oxo-5 alpha-steroid delta 4-dehydrogenase alpha-2)	264600	REa	Pseudovaginal perineoscrotal hypospadias (1)	
Chr.2	UGT1A1, UGT1, GNT1	P	UDP-glucuronosyltransferase-1 family, member 1	191740	REa	Crigler-Najjar syndrome, type I, 218800 (1); ?Gilbert syndrome, 143500 (1)	1(Ugt-1)
Chr.2	XDH	P	Xanthine dehydrogenase (xanthine oxidase)	278300	REb	Xanthinuria (1)	
3p26-p25	VHL	C	von Hippel-Lindau syndrome	193300	Fd, D, RE	von Hippel-Lindau syndrome (3); Renal cell carcinoma (1)	
3p25.3	PHS	L	Pallister-Hall syndrome	146510	Ch	?Pallister-Hall syndrome (2)	
3p25	XPC, XPCC	P	Xeroderma pigmentosum, complementation group C	278720	REa, A	Xeroderma pigmentosum, complementation group C (2)	
3p21.3	COL7A1	C	Collagen VII, alpha-1 polypeptide	120120	REa, A	Epidermolysis bullosa dystrophica, dominant, 131750 (3); Epidermolysis bullosa dystrophica, recessive, 226600 (3)	
3p14.3	TKT	P	Transketolase	277730	REa, A	{Wernicke-Korsakoff syndrome, susceptibility to} (1)	
3p14-qter	HRG	P	Histidine-rich glycoprotein	142640	REa	?Thrombophilia due to elevated HRG (1)	
3p11	PIT1	C	Pituitary-specific transcription factor Pit-1	173110	Fd, A	Pituitary hormone deficiency, combined (1)	16(Pit-1,dw)

Table 1 – contd

Location	*Symbol*	*Status*	*Title*	*MIM #*	*Method*	*Disorder*	*Mouse*
3q2	AKU	C	Alkaptonuria	203500	Fd, H	Alkaptonuria (2)	16(aku)
3q21-q24	HHC1, FHH, PCAR1	P	Hypocalciuric hypercalcemia-1 (parathyroid Ca(2+)-sensing receptor)	145980	Fd, REa	Hypercalcemia, hypocalciuric, familial (3); Hyperparathyroidism, neonatal 239200 (3)	
3q21-q24	RHO, RP4	C	Rhodopsin	180380	REa, A, Fd	Retinitis pigmentosa-4, autosomal dominant (3); Retinitis pigmentosa, autosomal recessive (3); Night blindness, congenital stationery (3)	6(Rho)
3q22-q23	BPES	C	Blepharophimosis, epicanthus inversus and ptosis	110100	Ch	Blepharophimosis, epicanthus inversus and ptosis (2)	
3q25-q26	SI	P	Sucrase-isomaltase	222900	REa, A, Fd	Sucrose intolerance (1)	3(Si-s)
3q26-qter	KNG	C	Kininogen	228960	Psh, A	[Kininogen deficiency] (3)	
3q27	BCL6	P	B-cell CLL/lymphoma-6	109565	Ch, A	Lymphoma, B-cell (2); Lymphoma, diffuse large cell (3)	
Chr.3	GBE1	P	Glycogen branching enzyme	232500	REa	Glycogen storage disease IV (1)	
4p16-p14	CDPR	L	Chondrodysplasia punctata, rhizomelic	215100	Ch	?Chondrodysplasia punctata, rhizomelic (2)	
4q21-q23	PKD2, PKD2B, PKD3	C	Polycystic kidney disease-2 (autosomal dominant)	173910	Fd	Polycystic kidney disease, adult, type II (2)	
4q26-q28	WBS, WMS	P	Williams-Beuren syndrome	194050	Ch	Williams-Beuren syndrome, type II (2)	
4q28	FGA	C	Fibrinogen, alpha polypeptide	134820	RE, REa, H, D, LD, A	Dysfibrinogenemia, alpha types (1); Amyloidosis, hereditary renal, 105200 (1)	
4q32-qter	ETFDH	P	Electron transfer flavoprotein: ubiquinone oxidoreductase	231675	REa, A	Glutaricacidemia type IIC (3)	
Chr.4	LAG5	P	Leukocyte antigen group 5	151450	S	Neutropenia, neonatal alloimmune (1)	
5q13.3	RASA, GAP	C	RAS p21 protein activator (GTPase activating protein)	139150	REa, A	Basal cell carcinoma (1)	13(Gap)
5q22-q33.3	CDGG1	P	Corneal dystrophy, Groenouw type I	121900	Fd	Corneal dystrophy, Groenouw type I (2); Corneal dystrophy, lattice type I, 122200 (2); Corneal dystrophy, combined granular/lattice type (2)	
5q23-q31	ITGA2, CD49B, BR	P	Integrin, alpha-2 (CD49B; alpha-2 subunit of VLA-2 receptor; platelet antigen Br)	192974	S, Psh, A	Thrombocytopenia, neonatal ? alloimmune (2); ?Glycoprotein Ia deficiency (2)	
5q31.1	IRF1	C	Interferon regulatory factor-1	147575	Fd, REa, A, REn, D	Macrocytic anemia refractory, of 5q-syndrome, 153550 (3); Myelodysplastic syndrome, preleukemic (3); Myelogenous leukemia, acute (3)	11(Irf1)
5q32-q33.1	GM2A	C	GM2 ganglioside activator protein	272750	S, REa, Psh	GM2-gangliosidosis, AB variant (1)	
5q33-q35	GLRA1, STHE	C	Glycine receptor, alpha 1	138491	Fd, R, A	Startle disease/hyperekplexia, 149400 (3)	11(spd)
5q34-q35	CRS2, MSX2	C	Craniosynostosis, type 2	123101	Fd, REa, A	Craniosynostosis, type 2 (3)	
6p23	SCA1	C	Spinal cerebellar ataxia-1	164400	F, Fd, A	Spinocerebellar ataxia-1 (3)	
6p21.3	AS, ANS	P	Ankylosing spondylitis	106300	Fd	Ankylosing spondylitis (2)	
6p21.3	HLA-DPB1	C	Major histocompatibility complex, class II, DP beta 1	142858	F, RE	{Beryllium disease, chronic, susceptibility to} (1)	
6p21.1-cen	RDS, RP7	C	Retinal degeneration,	179605	REa, A	Retinitis pigmentosa, peripherin-slow (peripherin) related (3); Retinitis punctata albescens (1); Macular dystrophy (1)	17(rds)
6p	PUJO	P	Pelviureteric junction obstruction	143400	F	Pelviureteric junction obstruction (2)	
6q21-q22.3	COL10A1	C	Collagen, type X, alpha-1 polypeptide	120110	REa, A	Metaphyseal chondrodysplasia, Schmid type (1)	10(Col10a1)
6q25.1	ESR, ER	C	Estrogen receptor	133430	REa, A	Breast cancer (1)	10(Esr)
6q26-q27	OVCS	P	Ovarian cancer, serous	167000	D	Ovarian cancer, serous (2)	
7p21.3-p21.2	CRS, CSO	C	Craniosynostosis, type I	123100	Ch	Craniosynostosis, type I (2)	
7p21	ACS3, SCS	C	Acrocephalosyndactyly-3 (Saethre-Chotzen syndrome)	101400	Fd, Ch	Saethre-Chotzen syndrome (2)	
7p15.1-p13	RP9	P	Retinitis pigmentosa-9	180104	Fd	Retinitis pigmentosa-9 (2)	
7q	RP10	P	Retinitis pigmentosa-10 (autosomal dominant)	180105	Fd	Retinitis pigmentosa-10 (2)	
7q11.2	CD36, GP4	P	CD36 antigen (collagen type I)	173510	A	[Macrothrombocytopenia] (1)	
7q11.2	ELN	C	Elastin	130160	REa, A, F, Fd	Supravalvar aortic stenosis, 185500 (3); Williams-Beuren syndrome, 194050 (3)	
7q31	CLD	P	Chloride diarrhea, congenital	214700	Fd	Chloride diarrhea, congenital (2)	
7q34-q35	TBXAS1	P	Thromboxane A sysnthasse 1 (platelet)	274180	A	Thromboxane synthase deficiency (2)	
7q35	CLCN1	P	Chloride channel-1, skeletal muscle	118425	H, REa, Fd	Myotonia congenita, recessive, 255700 (3); Myotonia congenita, dominant, 160800 (2)	6(adr, Clc-1)
7q36	HPE3, HLP3	C	Holoprosencephaly-3	142945	Ch, Fd	Holoprosencephaly, type 3 (2)	
8p21-p12	CLU, CLI, SGP2, TRPM2	C	Clusterin (complement lysis inhibitor, SP-40,40; sulfated glycoprotein 2; testosterone-repressed prostate message-2; apolipoprotein J)	185430	REa, REb, A, RE	{?Atherosclerosis, susceptibility to} (3)	14(Sgp2)
8p12-p11	WRN	C	Werner syndrome	277700	Fd	Werner syndrome (2)	

Table 1 – contd

Location	Symbol	Status	Title	MIM #	Method	Disorder	Mouse
8q	EBN2	P	Epilepsy, benign neonatal-2 (benign familial neonatal convulsions)	121201	Fd	Epilepsy, benign neonatal, type 2 (2)	
8q	AVED	P	Ataxia (Friedreich-like) with selective E deficiency	277460	Fd, LD	Friedreich-like ataxia, due to vitamin E deficiency (2)	
8q11	HYRC1	P	Hyperradiosensitivity of murine SCID mutation, complementing-1	202500	C	?Severe combined immunodeficiency, type I (1)	16(scid)
8q13-q21.1	CMT4A, CMT4	P	Charcot-Marie-Tooth neuropathy-4A (autosomal recessive) recessive)	214400	Fd	Charcot-Marie-Tooth disease, type IVA (2)	
8q13.3	BOR, BOS	C	Branchiootorenal syndrome	113650	Ch, Fd	Branchiootorenal dysplasia (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); Aldosteronism, glucocorticoid-remediable (1)	
8q21.1	PAF1, PMP35	C	Peroxisomal assembly factor-1 (35kD)	170993	RE	Zellweger syndrome-3 (3)	
8q24.11-q24.13	EXT1, EXT	P	Exostoses (multiple) 1	133700	Ch, Fd	Exostoses, multiple, type 1 (2)	
8q24.2-q24.3	TG	C	Thyroglobulin	188450	A, REa, REb	Hypothyroidism, hereditary congenital (1); Goiter, adolescent multinodular (1); Goiter, nonendemic, simple (1)	15(Tgn; cog)
9p23	TYRP, CAS2, CATB	C	Tyrosinase-related protein (catalase B; human homolog of 'brown' locus)	115501	Psh, REa, A	Albinism, brown, 203290 (1)	4(b;trp-1)
9p21	MLM, CMM2, MLM2	C	Melanoma	155601	D, Fd	Melanoma, cutaneous malignant (2)	
9p13-q11	CHH	P	Cartilage-hair hypoplasia	250250	Fd	Cartilage-hair hypoplasia (2)	
9q	ORW, HHT	P	Osler-Rendu-Weber syndrome) (hereditary hemorrhagic telangiectasia)	187300	Fd	Hereditary hemorrhagic telangiectasia (2)	
9q31-q33	DYS	P	Dysautonomia (Riley-Day syndrome, hereditary sensory autonomia neuropathy type III)	223900	Fd, LD	Dysautonomia, familial (2)	
9q31-q33	FCMD	P	Fukuyama type congenital muscular dystrophy	253800	Fd, LD	Fukuyama type congenital muscular dystrophy (2)	
9q32	AFDN	L	Acrofacial dysostosis, Nager type	154400	Ch	?Acrofacial dysostosis, Nager type (2)	
9q34.1	EPB72	C	Erythrocyte membrand protein band 7.2 (stomatin)	185000	REa, Ch, A	Stomatocytosis I (2)	
10q11.5	MEN2B, MEN3	C	Multiple endocrine neoplasia, type IIB	162300	Fd	Multiple endocrine neoplasia IIB (2)	
10q11.2	HSCR, MGC	C	Hirschsprung disease	142623	Ch, Fd, D	Megacolon (2)	?14(ls)
10q11.2	RET, MEN2A	C	RET transforming sequence; oncogene RET	164761	A, REn, Fd	Multiple endocrine neoplasia IIA, 171400 (3); Medullary thyroid carcinoma (3)	
10q11.2-q21	MBL, MBP1	C	Mannose-binding lectin	154545	REa, A, Fd	{Chronic infections, due to opsonin defect} (1)	
10q11	ERCC6, CKN2	P	Excision repair cross complementing rodent repair deficiency, complementation group 6	133540	A	Cockayne syndrome-2, late onset, 216410 (2)	
10q21-q22	DCOH, CADH	P	Dimerization cofactor of hepatic nuclear factor 1-alpha (TCF1) (4a-carbinolamine dehydratase)	126090	REa, H	Hyperphenylalaninemia due to 4a-carbinolamine dehydratase deficiency, 264070 (1)	10(Dcoh)
10q24.3	CYP17, P450C17	C	Cytochrome P450, subfamily XVII; steroid 17-alpha-hydroxylase	202110	REa, H, A	Adrenal hyperplasia, congenital, due to 17-alpha-hydroxylase deficiency (1)	19(Cyp17)
11pter-p13	AMPD3	P	Adenosine monophosphate deaminase 3 (isoform E)	102772	REa	[AMP deaminase deficiency, erythrocytic] (1)	
11p15.5	MTACR1, WT2	P	Multiple tumor associated chromosome region-1	194071	D	Wilms tumor, type 2 (2); Adrenocortical carcinoma, hereditary (2)	
11p13	CD59	C	Antigen CD59 (p18-20)	107271	REa, A, D	CD59 deficiency (3)	15(Ly-6)
11p13	RBTNL1, RBTN2, RHOM2, TTG2	P	Rhombotin-like 1	180385	REa, REc	Leukemia, acute, T-cell (2)	
11p13	ROM1, ROSP1	P	Rod outer segment membrane protein-1	180721	REa	Retinitis pigmentosa, autosomal dominant (1)	19(Rosp-1)
11p13-q13	CMH4	P	Cardiomyopathy, familial hypertrophic, 4	115197	Fd	Cardiomyopathy, familial hypertrophic, 4 (2)	
11p12-p11.12	GLNN	P	Galanin	137035	Psh, A	?Galanin deficiency syndrome (1)	19(Glnn)
11p12-p11.12	PFM	L	Parietal foramina	168500	Ch	?Parietal foramina (2)	
11p	USH1C	P	Usher syndrome-1C (autosomal recessive, severe)	276904	Fd	Usher syndrome, type 1C (2)	
11q	IDDM2	L	Insulin-dependent diabetes mellitus-2	125852	H, LD	?Insulin-dependent diabetes mellitus-2 (2)	
11q13	RT6	P	RT6 antigen (rat) homolog	180840	REa, A	?{Susceptibility to IDDM} (1)	7(Rt-6)
11q13	SMTN	P	Somatotrophinoma	102200	D	Somatotrophinoma (2)	
11q13	UQOR1	P	NADH:ubiquinone oxidoreductase, mitochondrial (51kD)	161015	REa	?Mitochondrial complex I deficiency, 252010 (1)	
11q13	VRNI	P	Vitreoretinopathy, neovascular inflammator	193235	Fd	Vitreoretinopathy, neovascular inflammatory (2)	
11q13.5	USH1B	P	Usher syndrome-1B (autosomal recessive, severe)	276903	Fd	Usher syndrome, type 1B (2)	
11q23	APOC3	C	Apolipoprotein C-III	107720	REa, RE, F	Hypertriglyceridemia (1)	
11q23	MLL, ALL1, HRX, TRX1	C	Myeloid/lymphoid, or mixed-lineage leukemia; trithorax (Drosophila) homolog	159555	Ch, RE	Leukemia, myeloid/lymphoid or mixed-lineage (2)	
11q23-q24	FLI1	C	Friend leukemia virus integration 1	193067	REa, Ch, A	Neuroepithelioma, 133450 (1)	
11q23.1	PORC	P	Porphyria, acute, Chester type	176010	Fd	Porphyria, Chester type (2)	

Table 1 – contd

Location	*Symbol*	*Status*	*Title*	*MIM #*	*Method*	*Disorder*	*Mouse*
12pter-p12	DRPLA	C	Dentatorubro-pallidoluysian atrophy	125370	Fd	Dentatorubro-pallidoluysian atrophy (3)	
12p13.3	F8VWF, VWF	C	Coagulation factor VIII VWF (von Willebrand factor)	193400	A, REa, REb, Fd	von Willebrand disease (1)	6(Vwf)
12p13.3-p11.2	ACLS	L	Acrocallosal syndrome	200990	Ch	?Acrocallosal syndrome (2)	
12p	AEM	P	Ataxia, episodic, with myokymia	160120	Fd	Ataxia, episodic, with myokymia (2)	
12q11-q13	KRT1, KRTA	C	Keratin-1	139350	H, REa, A	Epidermolytic hyperkeratosis, 113800 (3)	15(Krt-2)
12q11-q13	KRT5	P	Keratin-5	148040	A, Fd	Epidermolysis bullosa simplex, Dowling-Meara type, 131760 (3); Epidermolysis bullosa simplex, Koebner type, 131900 (3); ?Epidermolysis bullosa, Weber-Cockayne type, 131800 (3)	
12q12-q13	CD63, MLA1	P	Antigen CD63 (melanoma 1 antigen)	155740	REa, A	?Hermansky-Pudlak syndrome, 203300 (1)	
12q13	AQP2	P	Aquaporin 2 (collecting duct)	107777	A	?Diabetes insipidus nephrogenic, type II, 125800 (1)	
12q13.11-q13.2	COL2A1	C	Collagen, type II, alpha-1 polypeptide	120140	REa, A	Stickler syndrome, type I (3); SED congenita (3); Kniest dysplasia (1); Achondrogenesis- hypochondrogenesis, type II (1); Osteoarthrosis, precocious (3); Wagner syndrome, type II (1); SMED Strudwick type (1)	
12q14-qter	PPD	P	4-hydroxyphenylpyruvate dioxygenase	276710	REa	Tyrosinemia, type III (1)	
12q23-q24.1	DAR, DWD	P	Darier disease (keratosis follicularis)	124200	Fd	Darier disease (keratosis follicularis) (2)	
12q24	SCA2	C	Spinal cerebellar ataxia-2 (olivopontocerebellar ataxia-2)	183090	Fd	Spinocerebellar atrophy II (2)	
Chr.12	LYZ	P	Lysozyme	153450	REa	Amyloidosis, renal, 105200 (1)	
13q12-q13	DMDA1	P	Duchenne-like muscular dystrophy, autosomal recessive	253700	Fd	Muscular dystrophy, Duchenne-like, autosomal (2)	
13q14	DBM, D13S25	P	Disrupted in B-cell neoplasia	109543	D	Leukemia, chronic lymphocytic, B-cell (2)	
13q14.1-q14.2	RB1, OSRC	C	Retinoblastoma-1	180200	Ch, F, Fd	Retinoblastoma (3); Osteosarcoma, 259500 (2)	14(Rb-1)
13q14.3-q21.1	ATP7B, WND, WD	C	ATPase, Cu++ transporting, beta polypeptide	277900	F, Fd	Wilson disease (3)	
13q32-q34	ERCC5	C	Excision-repair, complementing defective, in Chinese hamster, number 5	133530	S, A	Xeroderma pigmentosum, group G (1)	
Chr.13	CPB2	P	Carboxypeptidase B2 (plasma)	212070	Psh	Carboxypeptidase B deficiency (1)	
14q	DRD	P	Dystonia, DOPA-responsive	128230	Fd	Dystonia, DOPA-responsive (2)	
14q	SPG3	P	Spastic paraplegia-3	182660	Fd	Spastic paraplegia-3 (2)	
14q11.1-q13	HPE4	L	Holoprosencephaly-4, semilobar	142946	Ch	?Holoprosencephaly-4 (2)	
14q11.2	TCRA	C	T-cell antigen receptor, alpha polypeptide	186880	H, REa, A, REn	Leukemia/lymphoma, T-cell (3)	14(Tcra)
14q21-q22	PYGL, PPYL	P	Phosphorylase, glycogen, liver	232700	REb	Glycogen storage disease VI (1)	12(Pygl)
14q24-qter	CAP1	L	Cataract, anterior polar, 1	115650	Ch	?Cataract, anterior polar, I (2)	
14q24.3	AD3	C	Alzheimer disease-3	104311	Fd	Alzheimer disease-3 (2)	
14q24.3-q31	MJD	P	Machado-Joseph disease	109150	Fd	Machado-Joseph disease (2)	
14q24.3-qter	SCA3	P	Spinocerebellar ataxia-3	183085	Fd	Spinocerebellar ataxia-3 (2)	
14q31	TSHR	C	Thyroid-stimulating hormone receptor	275200	REa, Fd, A	Hypothyroidism, nongoitrous, due to TSH resistance (1); Thyroid adenoma, hyperfunctioning (1); Graves disease, 275000 (1)	12(Tshr)
14q32	USH1A, USH1	P	Usher syndrome-1A	276900	Fd	Usher syndrome, type 1A (2)	
14q32.1	PCI, PLANH3, PAI3	C	Protein C inhibitor (plasminogen activator inhibitor-3)	227300	Psh, REn	Protein C inhibitor deficiency (2)	
15q11-q13	AHO2	L	Albright hereditary osteodystrophy-2	103581	D	?Albright hereditary osteodystrophy-2 (2)	
15q11.2-q12	P, PED, D15S12, OCA2	C	Pink-eyed dilution, homolog of	203200	D, REa, Fd	Albinism, oculocutaneous, type II (3)	7(p)
15q12	SNRPN	P	Small nuclear ribonucleoprotein	182279	REa, D	?Prader-Willi syndrome (1)	7(Snrpn)
15q2	CMH3	P	Cardiomyopathy, familial hypertrophic, 3 polypeptide N	115196	Fd	Cardiomyopathy, familial familial hypertrophic, 3 (2)	
15q21	DA3, CDA3	P	Dyserythropoietic anemia, type III	105600	Fd	Dyserythropoietic anemia, type III (2)	
15q26.1	BLM, BS	C	Bloom syndrome	210900	M, LD	Bloom syndrome (2)	
Chr.15	XPF	L	Xeroderma pigmentosum, complementation group F	278760	M	?Xeroderma pigmentosum, type F (2)	
16p13.3	CATM	P	Cataract, congenital, with microphthalmia	156850	Ch	Cataract, congenital, with microphthalmia (2)	
16p13	TSC2	C	Tuberous sclerosis-2	191092	Fd	Tuberous sclerosis-2 (2)	
16q21	BBS2	P	Bardet-Biedl syndrome-2	209900	Fd	Bardet-Biedl syndrome-2 (2)	
16q24.3	GALNS, MPS4A	C	N-acetylgalactosamine-6-sulfate sulfatase	253000	A, Psh	Mucopolysaccharidosis IVA (3)	
17p11.2	PMP22, CMT1A	C	Peripheral myelin protein-22	118220	Fd, D, A	Charcot-Marie-Tooth neuropathy, slow nerve conduction type Ia (2); Neuropathy, recurrent, with pressure palsies, 162500 (3); Dejerine-Sottas disease, PMP22 related, 145900 (3)	11(Tr)
17q11.2	NF1, VRNF, WSS	C	Neurofibromatosis, type 1 (neurofibromatosis, von Recklinghausen disease, Watson syndrome)	162200	Fd, EM, Ch, F	Neurofibromatosis, type I (3); Watson syndrome, 193520 (3)	

Table 1 – contd

Location	Symbol	Status	Title	MIM #	Method	Disorder	Mouse
17q11.2-q24	MHS2	P	Malignant hyperthermia susceptibility-2	154275	Fd	Malignant hyperthermia susceptibility-2, 145600 (2	
17q12-q21	EDH17B1, EDHB17A	C	Estradiol 17-beta-dehydrogenase-1	264300	A, REa	Pseudohermaphroditism, male, with gynecomastia (1); Polycystic ovarian disease (1)	
17q12-q21	EPPK, KRT9	C	Epidermolytic palmoplantar keratoderma	144200	Fd, REa	Epidermolytic palmoplantar keratoderma (3)	
17q21-q22	EPB3, EMPB3, AE1	C	Erythrocyte surface protein band 3 (anion exchanger-1)	109270	REa, RE, Fd, A	[Acanthocytosis, one form] (1); [Elliptocytosis, Malaysian-Melanesian type] (1); Spherocytosis, hereditary (3)	
17q21-q22	PENT, PNMT	C	Phenylethanolamine N-methyltransferase	171190	REa, Fd	?Hypertension, essential, 145500 (1)	
17q21.31-q22.05	COL1A1	C	Collagen I, alpha-1 polypeptide	120150	C, M, A, REa	Osteogenesis imperfecta, 4 clinical forms, 166200, 166210, 259420, 166220 (3); Ehlers-Danlos syndrome, type VIIA1, 130060 (3); Osteoporosis, idiopathic, 166710 (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); Thrombocytopenia, neonatal alloimmune (1)	
17q23	DCP1, ACE1	C	Dipeptidyl carboxypeptidase-1 (angiotensin I converting enzyme)	106180	A, H, Fd	{Myocardial infarction, susceptibility to} (3)	
17q23	GAA	C	Glucosidase, acid alpha-	232300	S, A, D, C	Glycogen storage disease, type II (1)	
17q24.3-q25.1	CMPD1, CMD1, SRA1	C	Campomelic dysplasia-1 (sex reversal, autosomal, 1)	211970	Ch	Campomelic dysplasia-1 (2)	?11(Ts)
17q25	RSS	L	Russell-Silver syndrome	270050	Ch	?Russell-Silver syndrome (2)	
18pter-q11	HPE1	L	Holoprosencephaly-1, alobar	236100	Ch	?Holoprosencephaly-1 (2)	
18p11.2	MC2R	P	Melanocortin-2 receptor (ACTH receptor)	202200	A	Glucocorticoid deficiency, due to ACTH unresponsiveness (1)	
18p	NPC	C	Niemann-Pick disease, type C	257220	Ch, H, Fd, M	Niemann-Pick disease, type C (2)	18(spm)
18q22-qter	MS1	L	Multiple sclerosis	126200	Fd, LD	{?Multiple sclerosis, susceptibility to} (2)	
Chr.18	FEO	P	Familial expansile osteolysis	174810	Fd	Familial expansile osteolysis (2)	
19p13.3	HHC2, FHH2	P	Hypocalciuric hypercalcemia, type 2	145981	Fd	Hypocalciuric hypercalcemia, type II (2)	
19p13.3-p13.2	EPOR	C	Erythropoietin receptor	133171	A, REa, H	[Erythrocytosis, familial], 133100 (2)	9(Epor)
19p13.2-q13.3	LPSA, D19S381E	P	Oncogene liposarcoma (DNA Segment, single copy, expressed, probes MC15, MC6)	164953	A	Liposarcoma (1)	
19cen-q13.2	AD2	C	Alzheimer disease-2 (late-onset)	104310	Fd	Alzheimer disease-2, late onset (2)	
19q12	CASIL	P	Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy	125310	Fd	Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy (2)	
19q12	MED	P	Multiple epiphyseal dysplasia	132400	Fd	Multiple epiphyseal dysplasia (2)	
19q12	PSACH	C	Pseudoachondroplastic dysplasia	177150	Fd	Pseudoachondroplastic dysplasia (2)	
19q12-q13	MHP	P	Migraine, familial hemiplegic	141500	Fd	Migraine, familial hemiplegic (2)	
19q13.1	RYR1, RYDR, MHS, CCO	C	Ryanodine receptor-1 (skeletal)	180901	A, Fd, H	Malignant hyperthermia susceptibility-1, 145600 (3); Central core disease, 117000 (3)	7(Ryr)
19q13.1-q13.2	AKT2	P	Murine thymoma viral (v-akt) homolog-2	164731	A	Ovarian carcinoma, 167000 (2)	
19q13.2-q13.3	PVS	C	Polio virus sensitivity	173850	S, A	{Polio, susceptibility to} (2)	9(Pvs)
19q13.3	GYS, GCGS	C	Glycogen synthase	138570	REa, A	{Non-insulin dependent diabetes mellitus, susceptibility to} (2)	
Chr.19	ETFB	P	Electron transfer flavoprotein, beta polypeptide	130410	REa	Glutaricaciduria, type IIB (2)	
20p12	BMP2, BMP2A	C	Bone morphogenetic protein-2	112261	H, REa, A	?Fibrodysplasia ossificans progressiva (1)	2(Bmp2a)
20q13.2	GNAS1, GNAS,	C	Guanine nucleotide-binding protein (G protein), alpha-stimulating activity polypeptide-1 GPSA	139320	REa, H, A, Fd	Pseudohypoparathyroidism, type Ia, 103580 (1); McCune-Albright polyostotic fibrous dysplasia, 174800 (1); Somatotrophinoma (1)	2(Gnas)
20q13.31	PCK1	C	Phosphoenolpyruvate carboxykinase-1 (soluble)	261680	REa, A, Fd	?Hypoglycemia due to PCK1) deficiency (1)	2(Pck1)
21q22.1	SOD1, ALS1	C	Superoxide dismutase-1, soluble	147450	S, D, Fd	Amytrophic lateral sclerosis, due to SOD1 deficiency, 105400 (3)	16(Sod-1)
22q12.2	NF2, ACN	C	Neurofibromatosis-2 (bilateral acoustic neuroma)	101000	RE, F, Ch, D, Fd	Neurofibromatosis, type 2 (3); Meningioma, NF2-related (3)	
22q13.1	CYP2D@, CYP2D, P450C2D	C	Cytochrome P450, subfamily IID	124030	F, Fd, Psh, A	{?Parkinsonism, susceptibility to} (1); Debrisoquine sensitivity (1)	15(Cyp2d)
Xpter-p22.32	GCFX, SS	P	Growth control factor, X-linked	312865	Fd	Short stature (2)	
Xpter-p22.2	CFND	L	Craniofrontonasal dysplasia	304110	Ch	?Craniofrontonasal dysplasia (2)	
Xp22.3	OASD	P	Ocular albinism and sensorineural deafness	300650	Fd	Ocular albinism with sensorineural deafness (2)	
Xp21.1	RP3	C	Retinitis pigmentosa-3 (X-linked recessive)	312610	Fd, D	Retinitis pigmentosa-3 (2)	
Xp21	SRS, MRSR	P	Snyder-Robinson X-linked mental retardation syndrome	309583	Fd	Mental retardation, Snyder-Robinson type (2)	
Xp11.4	NDP, ND	C	Norrie disease (pseudoglioma)	310600	Fd, D	Norrie disease (3); Exudative vitreoretinopathy, X-linked, 305390 (3)	
Xp11.23	MAOA	C	Monoamine oxidase A	309850	Fd, REa, D, A, REn	Brunner syndrome (3)	X(Maoa)
Xp11.2	SSRC	C	Sarcoma, synovial	312820	Ch, RE, A	Sarcoma, synovial (2)	

Table 1 – contd

Location	*Symbol*	*Status*	*Title*	*MIM #*	*Method*	*Disorder*	*Mouse*
Xp11	MRXA	L	Mental retardation, X-linked nonspecific, with aphasia	309545	Fd	?Mental retardation, X-linked nonspecific, with aphasia (2)	
Xq12-q13	ATP7A, MNK, MK	C	ATPase, Cu++ transporting, alpha polypeptide	309400	Fc, X/A, H	Menkes disease (2)	X(Mo)
Xq12-q21	JMS	P	Juberg-Marsidi syndrome	309590	Fd	Juberg-Marsidi syndrome (2)	
Xq12-q21.31	ATRX, ATR2	P	Alpha-thalassemia/mental retardation syndrome, X-linked	301040	Fd	Alpha-thalassemia/mental retardation syndrome, type 2 (2)	
Xq13	ASAT	L	Anemia, sideroblastic, with spinocerebellar ataxia	301310	Fd	?Anemia, sideroblastic, with spinocerebellar ataxia (2)	
Xq13	IL2RG, SCIDX1, SCIDX, IMD4	C	Interleukin-2 receptor, gamma	308380	Fd	Severe combined immunodeficiency, X-linked, 300400 (3)	
Xq13	PHKA1	C	Phosphorylase kinase, muscle, alpha polypeptide	311870	REa, A, REn	?Muscle glycogenosis (1)	X(Phka)
Xq13.1	GJB1, CX32 CMTX1	C	Gap junction protein, beta-1, 32kD (connexin 32)	304040	REa, Fd	Charcot-Marie-Tooth neuropathy, X-linked-1, dominant, 302800 (3)	X(Gjb1)
Xq13.1	RPS4X, CCG2, SCAR	C	Ribosomal protein S4, X-linked	312760	A, REa, REn	Turner syndrome (1)	X(Rps4x)
Xq21.1-q21.31	CPX	C	Cleft palate and/or ankyloglossia	303400	Fd, D	Cleft palate, X-linked (2)	
Xq21.3-q22	BTK, AGMX1, IMD1, XLA, AT	C	Bruton agammaglobulinemia tyrosine kinase	300300	H, Fd, A	Agammaglobulinemia, type 1, X-linked (3	X(xid, Btp)
Xq22	COL4A6	P	Collagen, type IV, alpha-6 polypeptide	303631	REn	Leiomatosis, diffuse (1); ?Alport syndrome, X-linked, type 2 (1)	
Xq22-q28	AIH3	L	Amelogenesis imperfecta-3, hypomaturation or hypoplastic type hypoplastic type	301201	Fd	?Amelogenesis imperfecta-3, hypoplastic type (2)	
Xq22.1	PIGA	P	Phosphatidylinositol glycan class A	311770	A	Paroxysmal nocturnal hemoglobinuria (3)	
Xq25-q26	HTX	P	Heterotaxy, X-linked visceral	306995	Fd	Heterotaxy, X-linked visceral (2)	
Xq25-q26.1	TAS	P	Thoracoabdominal syndrome	313850	Fd	Thoracoabdominal syndrome (2)	
Xq26	CD40LG, HIGM1, IGM	C	CD40 antigen ligand (hyper IgM syndrome)	308230	Fd, A, Psh	Immunodeficiency, X-linked, with hyper-IgM (3)	X(CD40l)
Xq26	GUST	P	Gustavson mental retardation syndrome (with microcephaly, optic atrophy, deafness)	309555	Fd	Gustavson syndrome (2)	
Xq26	SDYS, SGB	C	Simpson dysmorphia syndrome	312870	Fd	Simpson-Galabi-Behmel syndrome (2)	
Xq26	SHFD2	P	Split-hand/split-foot deformity, type 2	313350	Fd	Split-hand/split-foot deformity, type 2 (2)	
Xq28	DKC	C	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); Barth syndrome (2)	
Xq28	FRAXE, FMR2	P	Fragile site, X-linked, E	309548	Ch, REn	Mental retardation, X-linked, FRAXE type (3)	
Xq28	L1CAM, CAML1, HSAS1, HSAS	C	L1 cell adhesion molecule	308840	A, RE, H	Hydrocephalus due to aqueduct of Sylvius, 307000 (3)	X(L1cam)
Yp11.3	TDF, SRY	C	Testis determining factor (sex-determining region Y)	480000	Ch, Fd	Gonadal dysgenesis, XY type (1)	Y(Tdy, Sry)

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

Disorder	Location
[Acanthocytosis, one form] (1)	17q21-q22
Achondrogenesis-hypochondrogenesis, type II (1)	12q13.11-q13.2
?Acrocallosal syndrome (2)	12p13.3-p11.2
?Acrofacial dysostosis, Nager type (2)	9q32
Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency (1)	8q21
Adrenal hyperplasia, congenital, due to 17-alpha-hydroxylase deficiency (1)	10q24.3
Adrenocortical carcinoma, hereditary (2)	11p15.5
Agammaglobulinemia, type 1, X-linked (3)	Xq21.3-q22
Albinism, brown, 203290 (1)	9p23
Albinism, oculocutaneous, type II (3)	15q11.2-q12
?Albright hereditary osteodystrophy-2 (2)	15q11-q13
Aldosteronism, glucocorticoid-remediable (1)	8q21
Alkaptonuria (2)	3q2
Alpha-thalassemia/mental retardation syndrome, type 2 (2)	Xq12-q21.31
?Alport syndrome, X-linked, type 2 (1)	Xq22
Alveolar proteinosis, congenital, 265120 (1)	Chr.2
Alzheimer disease-2, late onset (2)	19cen-q13.2
Alzheimer disease-3 (2)	14q24.3
?Amelogenesis imperfecta-3, hypoplastic type (2)	Xq22-q28
[AMP deaminase deficiency, erythrocytic] (1)	11pter-p13
Amyloidosis, hereditary renal, 105200 (1)	4q28
Amyloidosis, renal, 105200 (1)	Chr.12
Amyotrophic lateral sclerosis, juvenile (2)	2q33-q35
Amytrophic lateral sclerosis, due to SOD1 deficiency, 105400 (3)	21q22.1
?Anemia, sideroblastic, with spinocerebellar ataxia (2)	Xq13
Aneurysm, familial, 100070 (1)	2q31
Ankylosing spondylitis (2)	6p21.3
Ataxia, episodic, with myokymia (2)	12p
{?Atherosclerosis, susceptibility to} (3)	8p21-p12
Bardet-Biedl syndrome-2 (2)	16q21
Barth syndrome (2)	Xq28
Basal cell carcinoma (1)	5q13.3
{Beryllium disease, chronic, susceptibility to} (1)	6p21.3
3-beta-hydroxysteroid dehydrogenase, type II, deficiency (3)	1p13.1
Blepharophimosis, epicanthus inversus and ptosis (2)	3q22-q23
Bloom syndrome (2)	15q26.1
Branchiootorenal dysplasia (2)	8q13.3
Breast cancer (1)	6q25.1
Brunner syndrome (3)	Xp11.23
Campomelic dysplasia-1 (2)	17q24.3-q25.1
Carboxypeptidase B deficiency (1)	Chr.13
?Cardiomyopathy (1)	2q35
Cardiomyopathy, familial hypertrophic, 2 (2)	1q3
Cardiomyopathy, familial hypertrophic, 3 (2)	15q2
Cardiomyopathy, familial hypertrophic, 4 (2)	11p13-q13
Cartilage-hair hypoplasia (2)	9p13-q11
?Cataract, anterior polar, I (2)	14q24-qter
Cataract, congenital, with microphthalmia (2)	16p13.3
CD3, zeta chain, deficiency (1)	1q23-q21.1
CD59 deficiency (3)	11p13
Central core disease, 117000 (3)	19q13.1
Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy (2)	19q12
Charcot-Marie-Tooth disease, type II (2)	1p36-p35
Charcot-Marie-Tooth disease, type IVA (2)	8q13-q21.1
Charcot-Marie-Tooth neuropathy, slow nerve conduction type Ia (2)	17p11.2
Charcot-Marie-Tooth neuropathy, slow nerve conduction type Ib, 118200 (3)	1q22
Charcot-Marie-Tooth neuropathy, X-linked-1, dominant, 302800 (3)	Xq13.1
Chloride diarrhea, congenital (2)	7q31
?Chondrodysplasia punctata, rhizomelic (2)	4p16-p14
{Chronic infections, due to opsonin defect} (1)	10q11.2-q21
Cleft palate, X-linked (2)	Xq21.1-q21.31
CMO II deficiency (1)	8q21
Cockayne syndrome-2, late onset, 216410 (2)	10q11
Colon cancer, familial, nonpolyposis type 1 (3)	2p16-p15
Corneal dystrophy, combined granular/lattice type (2)	5q22-q33.3
Corneal dystrophy, Groenouw type I (2)	5q22-q33.3
Corneal dystrophy, lattice type I, 122200 (2)	5q22-q33.3
?Craniofrontonasal dysplasia (2)	Xpter-p22.2
Craniosynostosis, type 2 (3)	5q34-q35
Craniosynostosis, type I (2)	7p21.3-p21.2
Crigler-Najjar syndrome, type I, 218800 (1)	Chr.2
?Cystinuria, 220100 (1)	2pter-q32.3
Darier disease (keratosis follicularis) (2)	12q23-q24.1
Debrisoquine sensitivity (1)	22q13.1
Dejerine-Sottas disease, myelin P(0)-related, 145900 (3)	1q22
Dejerine-Sottas disease, PMP22 related, 145900 (3)	17p11.2
Dentatorubro-pallidoluysian atrophy (3)	12pter-p12
?Diabetes insipidus nephrogenic, type II, 125800 (1)	12q13
Dysautonomia, familial (2)	9q31-q33
Dyserythropoietic anemia, type III (2)	15q21
Dysfibrinogenemia, alpha types (1)	4q28
Dyskeratosis congenita (2)	Xq28
Dystonia, DOPA-responsive (2)	14q
Ehlers-Danlos syndrome, type IV, 130050 (3)	2q31
Ehlers-Danlos syndrome, type VIIA1, 130060 (3)	17q21.31-q22.05
[Elliptocytosis, Malaysian-Melanesian type] (1)	17q21-q22
Endocardial fibroelastosis-2 (2)	Xq28
Epidermolysis bullosa dystrophica, dominant, 131750 (3)	3p21.3
Epidermolysis bullosa dystrophica, recessive, 226600 (3)	3p21.3
Epidermolysis bullosa simplex, Dowling-Meara type, 131760 (3)	12q11-q13
Epidermolysis bullosa simplex, Koebner type, 131900 (3)	12q11-q13
?Epidermolysis bullosa, Weber-Cockayne type, 131800 (3)	12q11-q13
Epidermolytic hyperkeratosis, 113800 (3)	12q11-q13
Epidermolytic palmoplantar keratoderma (3)	17q12-q21
Epilepsy, benign neonatal, type 2 (2)	8q
[Erythrocytosis, familial], 133100 (2)	19p13.3-p13.2
Exostoses, multiple, type 1 (2)	8q24.11-q24.13
Exudative vitreoretinopathy, X-linked, 305390 (3)	Xp11.4
Familial expansile osteolysis (2)	Chr.18
?Fibrodysplasia ossificans progressiva (1)	20p12
Fibromuscular dysplasia of arteries, 135580 (1)	2q31
Friedreich-like ataxia, due to vitamin E deficiency (2)	8q
Fukuyama type congenital muscular dystrophy (2)	9q31-q33
?Galanin deficiency syndrome (1)	11p12-p11.12
?Gilbert syndrome, 143500 (1)	Chr.2
Glanzmann thrombasthenia, type A (1)	17q21.32
Glaucoma, primary open angle, juvenile-onset (2)	1q21-q31
Glucocorticoid deficiency, due to ACTH unresponsiveness (1)	18p11.2
Glutaricacidemia type IIC (3)	4q32-qter
Glutaricaciduria, type IIB (2)	Chr.19
Glycogen storage disease IV (1)	Chr.3
Glycogen storage disease, type II (1)	17q23
Glycogen storage disease VI (1)	14q21-q22
?Glycoprotein Ia deficiency (2)	5q23-q31
GM2-gangliosidosis, AB variant (1)	5q32-q33.1
Goiter, adolescent multinodular (1)	8q24.2-q24.3
Goiter, nonendemic, simple (1)	8q24.2-q24.3
Gonadal dysgenesis, XY type (1)	Yp11.3
Graves disease, 275000 (1)	14q31
Gustavson syndrome (2)	Xq26
?Hartnup disease, 234500 (1)	2pter-q32.3
Hereditary hemorrhagic telangiectasia (2)	9q
?Hermansky-Pudlak syndrome, 203300 (1)	12q12-q13
Heterotaxy, X-linked visceral (2)	Xq25-q26
Holoprosencephaly, type 3 (2)	7q36
?Holoprosencephaly-1 (2)	18pter-q11
?Holoprosencephaly-4 (2)	14q11.1-q13
Homocystinuria due to MTHFR deficiency (3)	1p36.3
Hydrocephalus due to aqueduct of Sylvius, 307000 (3)	Xq28
Hydroxymethylglutaricaciduria (1)	1pter-p33
Hypercalcemia, hypocalciuric, familial (3)	3q21-q24
Hyperparathyroidism, neonatal 239200 (3)	3q21-q24
Hyperphenylalaninemia due to 4a-carbinolamine dehydratase deficiency, 264070 (1)	10q21-q22
?Hypertension, essential, 145500 (1)	17q21-q22
{Hypertension, essential, susceptibility to} (3)	1q42-q43
Hypertriglyceridemia (1)	11q23
Hypocalciuric hypercalcemia, type II (2)	19p13.3
?Hypoglycemia due to PCK1 deficiency (1)	20q13.31
Hypothyroidism, hereditary congenital (1)	8q24.2-q24.3
Hypothyroidism, nongoitrous, due to TSH resistance (1)	14q31
Immunodeficiency, X-linked, with hyper-IgM (3)	Xq26
?Insulin-dependent diabetes mellitus-2 (2)	11q
Juberg-Marsidi syndrome (2)	Xq12-q21
[Kininogen deficiency] (3)	3q26-qter
Kniest dysplasia (1)	12q13.11-q13.2
?Lactase deficiency, adult, 223100 (1)	2q21
?Lactase deficiency, congenital (1)	2q21
Lactic acidosis due to defect in iron-sulfur cluster of complex I (1)	2q33-q34
Leiomatosis, diffuse (1)	Xq22
Leukemia, acute, T-cell (2)	11p13
Leukemia, chronic lymphocytic, B-cell (2)	13q14
Leukemia, myeloid/lymphoid or mixed-lineage (2)	11q23
Leukemia/lymphoma, T-cell (3)	14q11.2
Liposarcoma (1)	19p13.2-q13.3
Lymphoma, B-cell (2)	3q27
Lymphoma, diffuse large cell (3)	3q27
Machado-Joseph disease (2)	14q24.3-q31
Macrocytic anemia refractory, of 5q- syndrome, 153550 (3)	5q31.1
[Macrothrombocytopenia] (1)	7q11.2
Macular dystrophy (1)	6p21.1-cen
Malignant hyperthermia susceptibility-1, 145600 (3)	19q13.1
Malignant hyperthermia susceptibility-2, 145600 (2)	17q11.2-q24
Malignant melanoma, cutaneous (2)	1p36
McCune-Albright polyostotic fibrous dysplasia, 174800 (1)	20q13.2
Medullary thyroid carcinoma (3)	10q11.2
Megacolon (2)	10q11.2
Melanoma, cutaneous malignant (2)	9p21

Table 2 – contd

Disorder	*Location*
Meningioma, NF2-related (3)	22q12.2
Menkes disease (2)	Xq12-q13
Mental retardation, Snyder-Robinson type (2)	Xp21
Mental retardation, X-linked, FRAXE type (3)	Xq28
?Mental retardation, X-linked nonspecific, with aphasia (2)	Xp11
Metaphyseal chondrodysplasia, Schmid type (1)	6q21-q22.3
Migraine, familial hemiplegic (2)	19q12-q13
?Mitochondrial complex I deficiency, 252010 (1)	11q13
Mucopolysaccharidosis IVA (3)	16q24.3
Multiple endocrine neoplasia IIA, 171400 (3)	10q11.2
Multiple endocrine neoplasia IIB (2)	10q11.5
Multiple epiphyseal dysplasia (2)	19q12
{?Multiple sclerosis, susceptibility to} (2)	18q22-qter
?Muscle glycogenosis (1)	Xq13
Muscular dystrophy, Duchenne-like, autosomal (2)	13q12-q13
Myelodysplastic syndrome, preleukemic (3)	5q31.1
Myelogenous leukemia, acute (3)	5q31.1
{Myocardial infarction, susceptibility to} (3)	17q23
?Myopathy, desminopathic (1)	2q35
Myotonia congenita, dominant, 160800 (2)	7q35
Myotonia congenita, recessive, 255700 (3)	7q35
Nephronophthisis, juvenile (2)	2p23-cen
Neuroepithelioma, 133450 (1)	11q23-q24
Neurofibromatosis, type 2 (3)	22q12.2
Neurofibromatosis, type I (3)	17q11.2
Neuropathy, recurrent, with pressure palsies, 162500 (3)	17p11.2
Neutropenia, neonatal alloimmune (1)	Chr.4
Niemann-Pick disease, type C (2)	18p
Night blindness, congenital stationery (3)	3q21-q24
{Non-insulin dependent diabetes mellitus, susceptibility to} (2)	19q13.3
Norrie disease (3)	Xp11.4
Ocular albinism with sensorineural deafness (2)	Xp22.3
Osteoarthrosis, precocious (3)	12q13.11-q13.2
Osteogenesis imperfecta, 4 clinical forms, 166200, 166210, 259420, 166220 (3)	17q21.31-q22.05
Osteoporosis, idiopathic, 166710 (3)	17q21.31-q22.05
Osteosarcoma, 259500 (2)	13q14.1-q14.2
Ovarian cancer, serous (2)	6q26-q27
Ovarian carcinoma, 167000 (2)	19q13.1-q13.2
?Pallister-Hall syndrome (2)	3p25.3
?Parietal foramina (2)	11p12-p11.12
{?Parkinsonism, susceptibility to} (1)	22q13.1
Paroxysmal nocturnal hemoglobinuria (3)	Xq22.1
Pelviureteric junction obstruction (2)	6p
Pheochromocytoma (2)	1p
Pituitary hormone deficiency, combined (1)	3p11
Platelet alpha/delta storage pool deficiency (1)	1q23-q25
{Polio, susceptibility to} (2)	19q13.2-q13.3
Polycystic kidney disease, adult, type II (2)	4q21-q23
Polycystic ovarian disease (1)	17q12-q21
Porphyria, Chester type (2)	11q23.1
?Prader-Willi syndrome (1)	15q12
Precocious puberty, male, 176410 (1)	2p21
{Preeclampsia, susceptibility to} (3)	1q42-q43
Protein C inhibitor deficiency (2)	14q32.1
Pseudoachondroplastic dysplasia (2)	19q12
Pseudohermaphroditism, male, with gynecomastia (1)	17q12-q21
Pseudohypoparathyroidism, type Ia, 103580 (1)	20q13.2
Pseudovaginal perineoscrotal hypospadias (1)	Chr.2
Renal cell carcinoma (1)	3p26-p25
Retinitis pigmentosa, autosomal dominant (1)	11p13
Retinitis pigmentosa, autosomal recessive (3)	3q21-q24
Retinitis pigmentosa, peripherin-related (3)	6p21.1-cen
Retinitis pigmentosa-10 (2)	7q
Retinitis pigmentosa-3 (2)	Xp21.1
Retinitis pigmentosa-4, autosomal dominant (3)	3q21-q24
Retinitis pigmentosa-9 (2)	7p15.1-p13
Retinitis punctata albescens (1)	6p21.1-cen
Retinoblastoma (3)	13q14.1-q14.2
Rhabdomyosarcoma, alveolar (3)	2q35
?Russell-Silver syndrome (2)	17q25
Saethre-Chotzen syndrome (2)	7p21
Sarcoma, synovial (2)	Xp11.2
SED congenita (3)	12q13.11-q13.2
?Severe combined immunodeficiency, type I (1)	8q11
Severe combined immunodeficiency, X-linked, 300400 (3)	Xq13
Short stature (2)	Xpter-p22.32
Simpson-Galabi-Behmel syndrome (2)	Xq26
SMED Strudwick type (1)	12q13.11-q13.2
Somatotrophinoma (1)	20q13.2
Somatotrophinoma (2)	11q13
Spastic paraplegia-3 (2)	14q
Spherocytosis, hereditary (3)	17q21-q22
Spinocerebellar ataxia-1 (3)	6p23
Spinocerebellar ataxia-3 (2)	14q24.3-qter
Spinocerebellar atrophy II (2)	12q24
Split-hand/split-foot deformity, type 2 (2)	Xq26
Stargardt macular dystrophy (2)	1p21-p13
Startle disease/hyperekplexia, 149400 (3)	5q33-q35
Stickler syndrome, type I (3)	12q13.11-q13.2
Stomatocytosis I (2)	9q34.1
Sucrose intolerance (1)	3q25-q26
Supravalvar aortic stenosis, 185500 (3)	7q11.2
?{Susceptibility to IDDM} (1)	11q13
Thoracoabdominal syndrome (2)	Xq25-q26.1
Thrombocytopenia, neonatal alloimmune (1)	17q21.32
Thrombocytopenia, neonatal alloimmune (2)	5q23-q31
?Thrombophilia due to elevated HRG (1)	3p14-qter
Thromboxane synthase deficiency (2)	7q34-q35
Thyroid adenoma, hyperfunctioning (1)	14q31
{?Tuberculosis, susceptibility to} (2)	2q
Tuberous sclerosis-2 (2)	16p13
Turner syndrome (1)	Xq13.1
Tyrosinemia, type III (1)	12q14-qter
Urate oxidase deficiency (1)	1p22
Usher syndrome, type 1A (2)	14q32
Usher syndrome, type 1B (2)	11q13.5
Usher syndrome, type 1C (2)	11p
Vitreoretinopathy, neovascular inflammatory (2)	11q13
von Hippel-Lindau syndrome (3)	3p26-p25
von Willebrand disease (1)	12p13.3
Waardenburg syndrome, type I (3)	2q35
Waardenburg syndrome, type III, 148820 (3)	2q35
Wagner syndrome, type II (1)	12q13.11-q13.2
Watson syndrome, 193520 (3)	17q11.2
Werner syndrome (2)	8p12-p11
{Wernicke-Korsakoff syndrome, susceptibility to} (1)	3p14.3
Williams-Beuren syndrome, 194050 (3)	7q11.2
Williams-Beuren syndrome, type II (2)	4q26-q28
Wilms tumor, type 2 (2)	11p15.5
Wilson disease (3)	13q14.3-q21.1
Wrinkly skin syndrome (2)	2q32
Xanthinuria (1)	Chr.2
Xeroderma pigmentosum, complementation group C (2)	3p25
Xeroderma pigmentosum, group G (1)	13q32-q34
?Xeroderma pigmentosum, type F (2)	Chr.15
Zellweger syndrome-2 (1)	1p22-p21
Zellweger syndrome-3 (3)	8q21.1