The Human Gene Map 15 November 1983

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The Morbid Anatomy of the Human Genome,

see Figure 12, page 122–123.

Please call my attention
to errors of omission
or commission. V.A.M.

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November 15, 1983

HUMAN GENE MAP

The following information, updated periodically since 1973, is based in large part on seven International Workshops on Human Gene Mapping. The first was organized by Dr. Frank Ruddle and held in New Haven in June 1973. The second, known as the Rotterdam Conference, was organized by Dr. Dirk Bootsma and held in The Netherlands in July 1974. The third, organized by Dr. Victor McKusick, was held in Baltimore in October 1975. The fourth, organized by Dr. John Hamerton, was held in Winnipeg in August 1977. The fifth, organized by Dr. John Evans, was held in Edinburgh in July 1979. The sixth, organized by Dr. Kare Berg, was held in Oslo in June-July, 1981. The seventh, organized by Dr. Robert Sparkes, was held in Los Angeles in August, 1983. The first six were sponsored exclusively by the National Foundation-March of Dimes (now March of Dimes Birth Defects Foundation) which publishes the proceedings as part of its Birth Defects: Original Article Series. The proceedings also appear in Cytogenetics and Cell Genetics.

	BD:OAS	Cytogenet. Cell Genet.
HGM-1	X(3):1-216, 1974	13:1-216, 1974
HGM-2	XI(3):1-310, 1975	14:162-480, 1975
HGM-3	XII(7):1-452, 1976	16:1-452, 1976
HGM-4	XIV(4):1-730, 1978	22:1-730, 1978
HGM-5	XV(11):1-236, 1979	25:2-236, 1979
HGM-6	XVIII(2):1-343,1982	32:1-343, 1982
HGM-7	In press	In press

The methods for mapping genes are symbolized as follows:

1. F - linkage study in families; e.g., linkage of ABO blood group and nail-patella syndrome. (When a chromosomal heteromorphism or rearrangement is one trait, Fc is used; e.g., Duffy blood group locus on chromosome 1. When a DNA polymorphism is one trait, Fd may be used; e.g., Huntington disease on chromosome 4.)

2. S - "segregation" (cosegregation) of cellular traits and chromosomes (or segments of chromosomes) in particular clones from somatic cell hybrids, e.g., thymidine kinase to chromosome 17.

3. M - Microcell mediated gene transfer (MMGT), e.g., a collagen gene (COL1A1) to chromosome 17.

4. C - chromosome mediated gene transfer (CMGT), e.g., cotransfer of galactokinase and thymidine kinase. (In conjunction with this approach fluorescence-activated flow sorting can be used for transfer of specific chromosomes.)

5. R - irradiation of cells followed by "rescue" through fusion with nonirradiated (nonhuman) cell (Goss-Harris method of radiation-induced gene segregation); e.g., order of genes on Xq. (Also called cotransference. The complement of cotransference = recombination.)

6. A - in situ DNA-RNA or DNA-DNA annealing ("hybridization"), e.g., ribosomal RNA genes to acrocentric chromosomes; kappa light chain genes to chromosome 2.

7. HS - DNA/cDNA molecular hybridization in solution ("Cot analysis"), e.g., assignment of Hb beta to chr. 11 in derivative hybrid cells.

8. RE - Restriction endonuclease techniques, e.g., fine structure map of non-alpha globin (NAG) region on (beta-globin cluster, HBBC) 11p; physical linkage of 3 fibrinogen genes (on 4q) and APOA1 and APOC3 (on 11p).
   a. Combined with somatic cell hybridization, e.g., NAG (HBBC) to 11p
   b. Combined with chromosome sorting, e.g., insulin to 11p.

9. D - deletion 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 triplicate state of part or all of a chromosome with 50% more gene product). Examples: acid phosphatase-1 to chromosome 2; glutathione reductase to chromosome 8.

10. AAS - deductions from the amino acid sequence of proteins; e.g., linkage of delta and beta hemoglobin loci from study of hemoglobin Lepore. (Includes deductions of hybrid protein structure by monoclonal antibodies, e.g., close linkage of MN and Ss from study of Lepore-like MNSs blood group antigen.)

11. LD - linkage disequilibrium, e.g., beta and delta globin genes (HBB, HBD).

12. V - induction of microscopically evident chromosomal change by adenovirus (probably represents change comparable to "puffing" in insects; accompanied by activation of kinases), e.g., adenovirus 12 changes on chr. 1 and 17.

13. Ch - chromosomal change associated with particular phenotype and not proved to represent linkage (Fc), deletion (D) or virus effect (V); e.g., loss of 13q14 band in some cases of retinoblastoma. ("Fragile sites," observed in cultured cells with or without folate-deficient medium or BrdV treatment, falls into this class of method, e.g., fragile site at Xq27 in one form of X-linked mental retardation. Fragile sites are useful as markers in family linkage studies, e.g., FS16q22 and haptoglobin.)

14. OT - ovarian teratoma (centromere mapping), e.g., PGM3 and centromere of chr. 6.

15. EM - exclusion mapping, i.e., 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; e.g., support for assignment of MNSs to 4q.

16. H - based on presumed homology; e.g., assignment of LDHC to 12p.

The certainty with which assignment of loci to chromosomes or the linkage between two loci has been established has been graded into the following classes:

C = confirmed - observed in at least two laboratories or in several families (not used in the following lists but can be assumed when one of the other symbols is not given).

P = provisional - based on evidence from one laboratory or one family.

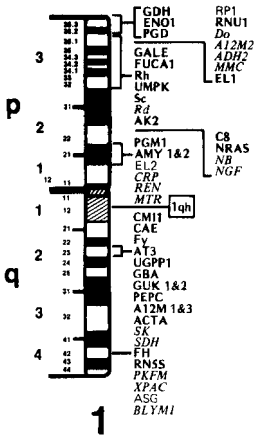
I = inconsistent - results of different laboratories disagree.

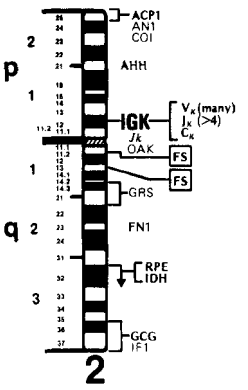
L = limbo group - evidence not as strong as provisional, but included for heuristic reasons. (Same as "tentative" in other terminology.)

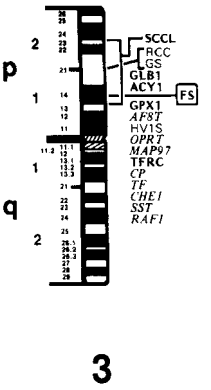
The five digit numbers given here are entry numbers in McKusick's Mendelian Inheritance in Man (6th ed., 1983, Johns Hopkins Univ. Press) with additions. Documentation on linkages and assignments is referenced in the entries indicated. The banding numerology is that of the Paris Conference (1971) with the 1975 Supplement (updated in the 1978 report of the Standing Committee on Human Cytogenetic Nomenclature, BD: OAS XIV (8): 1-92, 1978 and Cytogenet. Cell Genet. 21:313-404, 1978). Also see Yunis, Science 191:1268, 1976; Francke and Oliver, Hum. Genet. 45: 137-165, 1978. For report on high-resolution banding, see ISCN (1981): BD:OAS XVII (5):1-32, 1981 and Cytogenet. Cell Genet. 31:5-32, 1981.

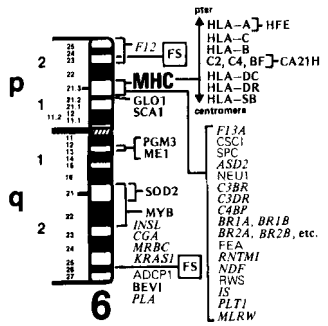
TABLE I
Number of loci assigned at each of the seven human gene mapping workshops

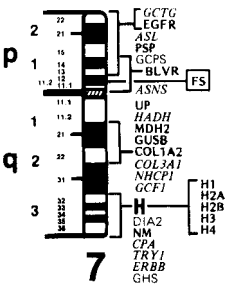
Conference	Number of autosomal assignments*				X chromosome assignments	
	Confirmed	Provisional	Inconsistent (inc. limbo)	Total	Confirmed	In Limbo
New Haven (1973)	31	28	5	64	88	67
Rotterdam (1974)	48	32	6	86	91	70
Baltimore (1975)	72	46	7	125	95	80
Winnipeg (1977)	83	82	11	176	102	96
Edinburgh (1979)	123	87	20	230	112	101
Oslo (1981)	180	120	45	345	116	118
Los Angeles (1983)	247	161	80	488	118	136
(+ 95 single copy DNA segments, 41 antigens, and 23 O'Farrell protein spots, all of unknown function, and 19 fragile sites)					(+76 DNA segments, 3 spots, 6 antigens, 2 FS)	

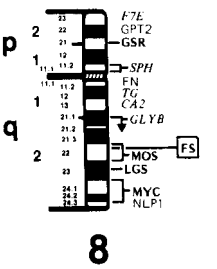

GDH
ENO1
PGD
GALE
FUCA1
Rh
UMPK
Sc
Rd
AK2
PGM1
AMY 1&2
EL2
CRP
REN
MTR
1qh
CMI1
CAE
Fy
AT3
UGPP1
GBA
GUK 1&2
PEPC
A12M 1&3
ACTA
SK
SDH
FH
RN5S
PKFM
XPAC
ASG
BLYM1
RP1
RNU1
Do
A12M2
ADH2
MMC
EL1
C8
NRAS
NB
NGF
p
q
1

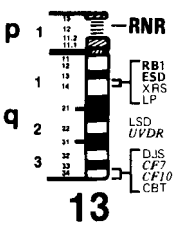

ACP1
AN1
COI
AHH
IGK
Jk
OAK
GRS
FN1
RPE
IDH
GCG
IF1
Vκ (many)
Jκ (>4)
Cκ
FS
p
q
2

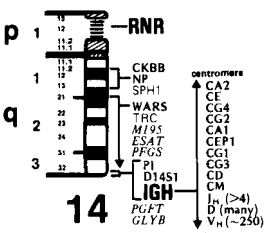

SCCL
RCC
LGS
GLB1
ACY1
GPX1
AF8T
HV1S
OPRT
MAP97
TFRC
CP
TF
CHE1
SST
RAF1
FS
p
q
3

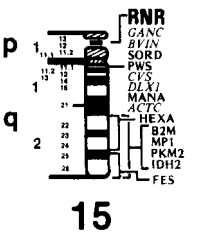

ptter
HLA–A
HLA–C
HLA–B
C2, C4, BF
HLA–DC
HLA–DR
HLA–SB
centromere
HFE
CA21H
F12
MHC
GLO1
SCA1
PGM3
ME1
SOD2
MYB
INSL
CGA
MRBC
KRAS1
ADCP1
BEVI
PLA
F13A
CSC1
SPC
ASD2
NEU1
C3BR
C3DR
C4BP
BR1A, BR1B
BR2A, BR2B, etc.
FEA
RNTM1
NDF
RWS
IS
PLT1
MLRW
FS
p
q
6

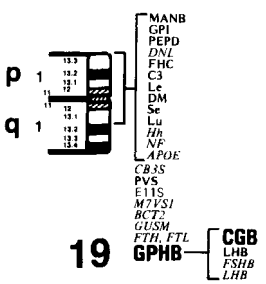

GCTG
EGFR
ASL
PSP
GCPS
BLVR
ASNS
UP
HADH
MDH2
GUSB
COL1A2
COL3A1
NHCP1
GCF1
H
DIA2
NM
CPA
TRY1
ERBB
GHS
H1
H2A
H2B
H3
H4
FS
p
q
7

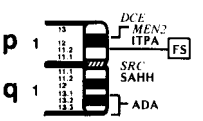

F7E
GPT2
GSR
SPH
FN
TG
CA2
GLYB
MOS
LGS
MYC
NLP1
FS
p
q
8


RNR
RB1
ESD
XRS
LP
LSD
UVDR
DJS
CF7
CF10
CBT
p
q
13


RNR
CKBB
NP
SPH1
WARS
TRC
M195
ESAT
PFGS
PI
D14S1
IGH
PGFT
GLYB
centromere
CA2
CE
CG4
CG2
CA1
CEP1
CG1
CG3
CD
CM
Jμ (>4)
D (many)
Vμ (~250)
q ter
p
q
14


RNR
GANC
BYIN
SORD
PWS
CYS
DLX1
MANA
ACTC
HEXA
B2M
MP1
PKM2
IDH2
FES
p
q
15


MANB
GPI
PEPD
DNL
FHC
C3
Le
DM
Se
Lu
Hh
NF
APOE
CB3S
PVS
E11S
M7TS1
BCT2
GUSM
FTH, FTL
GPHB
CGB
LHB
FSHB
LHB
p
q
19


DCE
MEN2
ITPA
SRC
SAHH
ADA
FS
p
q
20

A confirmed assignment ENO1
A provisional assignment . . . . . *DHPR*
Assignment "in limbo" (tentative, inconsistent) . . . . . Do
Gene cluster . . . . . **MHC**

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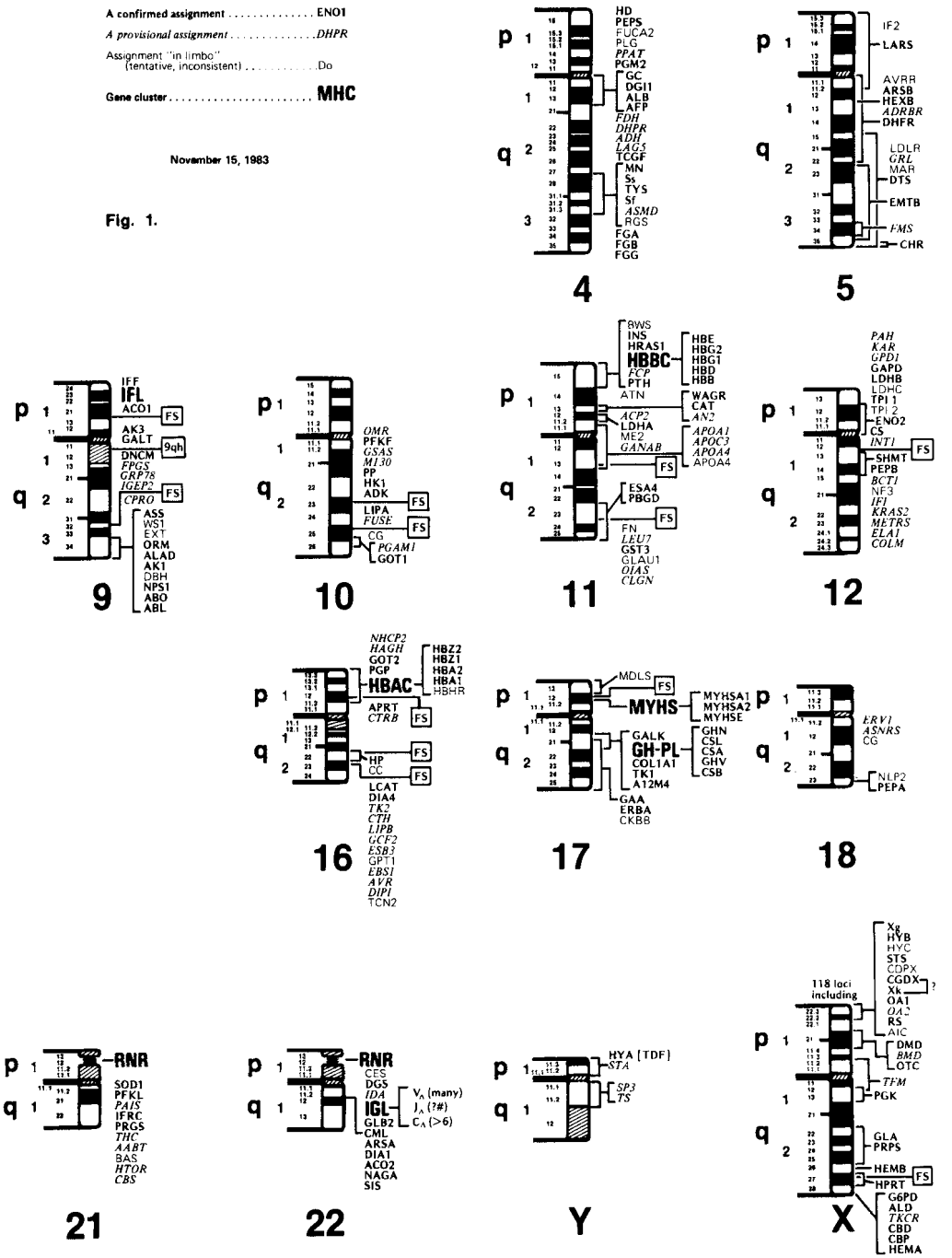
Fig. 1.



Key for Fig. 1

These symbols conform, for the most part, to those recommended by the Committee on Standardized Human Gene Nomenclature chaired by T. B. Shows and P. J. McAlpine, and O. J. Miller (HGM-7, Los Angeles, 1983). The rules include: 1. Only caps, except in blood group symbols. 2. No hyphens, except in HLA. 3. No more than 4 or 5 letters or numbers. 4. Chromosome number in symbols only for DNA segments, e.g., D14S1. S= segment. The last number refers to sequence of delineation on particular chromsome. "Z" replaces "S" if the DNA fragment contains repetitive sequences. (Only a few DNA segments that have been mapped are shown, for illustrative purposes.) 5. Cytoplasmic (cytosolic, or soluble) isozyme = "1", mitochondrial isozyme = "2". 6. Only arabic numbers, e.g. HpaI = HPA1. 7. Cell surface antigens symbolized S followed by sequential number (in order of delineation). (In addition to "anonymous," i.e. function unknown, DNA segments and cell surface antigens, some peptides identified by O'Farrell 2-D electrophoresis and some antigens identifed by monoclonal antibodies have been mapped. These, with symbols beginning P and M, respectively, have not been included here except when they have been used for the assignment of other loci.)

The location given is the SRO (shortest region of overlap) based on a collation of studies of chromosomal rearrangements. The location in parentheses is a shorter interval which takes gene order data into account, or is an assignment made indirectly through linkage to another assigned locus. A question mark precedes assignments which are inconsistent or "in limbo."

A12M1	= Adenovirus-12 chromosome modification site-1C - 1q42-43 (V)
A12M2	= Adenovirus-12 chromosome modification site-1A - 1p36 (V)
A12M3	= Adenovirus-12 chromosome modification site-1B - 1q21 (V)
A12M4	= Adenovirus-12 chromosome modification site-17 - 17q21-q22 (V)
AABT	= Beta-amino acids, renal transport of - ?chr. 21 (D)
ABL	= Onc gene: Abelson strain of murine leukemia virus - 9q34 (S)
ABO	= ABO blood group - 9q34 (F)
ACMA	= Actin, cardiac muscle alpha - 15q11-qter (REa)
ACO1	= Aconitase, soluble - 9p22-p13 (S)
ACO2	= Aconitase, mitochondrial - 22q11-q13 (S)
ACP1	= Acid phosphatase-1 - 2p23 or 2p25 (D, S)
ACP2	= Acid phosphatase-2 - 11p12-cen (S)
ACTA	= Actin, skeletal muscle alpha - 1p12-qter (REa)
ACY1	= Aminoacylase-1 - 3pter-q13 (S)
ADA	= Adenosine deaminase - 20q13-qter (S, D)
ADCP1	= Adenosine deaminase complexing protein-1 - ?chr. 6 (S)
ADCP2	= Adenosine deaminase complexing protein-2 - 2p23-2q32 (S)
ADH	= Alcohol dehydrogenase, class 1 - chr. 4 (REa)
ADH2	= Alcohol dehydrogenase-2 - 1pter-1p23 (S)
ADK	= Adenosine kinase - 10q11-q24 (S, D, EM)
ADRBR	= Beta-adrenergic receptor - chr. 5 (S)
AF8T	= Temperature-sensitive (AF8) complement - chr. 3 (S)
AFP	= Alpha-fetoprotein - 4q11-q13 (H, A)
AG(HBAC)	= ALPHA GLOBIN GENE CLUSTER - 16p12-pter (S, HS, A, D)
AHCY	= SAHH (q.v.)
AHH	= Aryl hydrocarbon hydroxylase - 2p (S)
AIC	= Aicardi syndrome - ?Xp22 (Ch)
AK1	= Adenylate kinase-1 (soluble) - 9q34 (F, S, D)
AK2	= Adenylate kinase-2 (mitochondrial) - 1p34 (S, F, R)
AK3	= Adenylate kinase-3 (mitochondrial) - 9p24-p13 (S)
AKE	= Acrokeratoelastoidosis - ?2p (F)
ALAD	= Delta-aminolevulinate dehydratase - (9q34; linked to ABO) (F)
ALB	= Albumin - 4q11-q13 (F, A, REa)
ALD	= Adrenoleukodystrophy - (Xq28; linked to G6PD) (F)
AMY1	= Amylase, salivary - 1p21 (F, A, REa)
AMY2	= Amylase, pancreatic - 1p21 (F, A, REa)
AN1	= Aniridia, type 1 (chr. 2; ?linked to ACP1) (F)
AN2	= Aniridia-2 - ?11p13 (?separate from WAGR) (Ch)
APOA1	= Apolipoprotein A-I - 11p11-q13 (REa, RE, F)
APOC2	= Apolipoprotein C-II - ?11p11-q13 (F)
APOC3	= Apolipoprotein C-III - 11p11-q13 (REa, RE, F)
APOA4	= Apolipoprotein A-IV - ?11p11-p13 (F)
APOE	= Apolipoprotein E - chr. 19 (F; linked to C3)
APRT	= Adenine phosphoribosyltransferase - 16q12-q22 (S, D)
ARSA	= Arylsulfatase A - 22q133l-qter (S)

ARSB = Arylsulfatase B - chr. 5 (S)
ASD2 = Atrial septal defect, secundum type (chr. 6; linked to HLA) (F)
ASG = Aspermiogenesis factor - ?1p13 or 1q25 (Ch)
ASL = Argininosuccinate lyase - 7p21-q22 (S)
ASMD = Anterior segment mesenchymal dysgenesis (chr. 4; linked to MNSs) (F)
ASNRS (NARS) = Asparaginyl-tRNA synthetase - chr. 18 (REa)
ASNS = Asparagine synthetase - 7p11-q11 (S)
ASS = Argininosuccinate synthetase - 9q34 (S, D)
ASSP2 = Argininosuccinate synthetase pseudogene-2 - chr. 6 (REa)
AT3 = Antithrombin III - 1q23.1-q23.9 (F, D, A, REa)
ATN = Tyrosinase negative albinism - (?11p; linked to NAG in mouse) (H)
ATPM = OMR (q.v.)
AVR(IFR) = Antiviral state regulator - chr. 16 (D)
AVRR = Antiviral state repressor regulator - ?5p (S)

B2M = Beta-2-microglobulin - 15q21-q22 (S, H)
BA2R = BALB/c 3T3 ts2 temperature sensitivity complementing - Xq13-Xq27 (S)
BAS = Beta-adrenergic stimulation, response to - ?chr. 21 (D)
BCT1 = Branched chain amino acid transferase-1 - 12pter-q12 (S)
BCT2 = Branched chain amino acid transferase-2 - chr. 19 (S)
BEVI = Baboon M7 virus infection - chr. 6 (S)
BF = Properdin factor B - 6p21.3 (in MHC) (F)
BLVR = Biliverdin reductase - 7p14-cen (S)
BLYM = Oncogene BLYM: chicken bursal lymphoma - chr. 1 (REa)
BR1A, BR1B, etc. = Blastogenic response to specific synthetic polypeptides (Ir homologs) - 6p21.3 (F)
BVIN = BALB virus induction, N-tropic - chr. 15 (S)
BVIX = BALB virus induction, xenotropic - chr. 11 (S)
BWS = *Beckwith-Wiedemann syndrome* - ?11p13-p15 (Ch)

C2 = Complement component-2 - 6p21.3 (in MHC) (F)
C3 = Complement component-3 - 19pter-q13.2 (S)
C3BR = Receptor for C3b - 6p21.3 (in MHC) (S)
C3DR = Receptor for C3d - 6p21.3 (in MHC) (S)
C4BP = Complement component-4 binding protein - ?chr. 6 (H)
C4F(C4A) = Complement component-4 fast - chr. 6 (in MHC) (F)
C4S(C4B) = Complement component-4 slow - chr. 6 (in MHC) (F)
C8 = Complement component-8 - 1p (F)
CA2 = Carbonic anhydrase II - chr. 8 (S)
CAE = Cataract, zonular pulverulent (1cen-q21); linked to Fy) (F)
CAH1(CA21H) = Congenital adrenal hyperplasia-1 (21-hydroxylase deficiency) - (6p21) (F)
CAT = Catalase - 11p13 (S)
CB3S = Coxsackie B3 virus susceptibility - chr. 19 (S)
CBBM = Colorblindness, blue monochromatic - ?Xq28 (F)
CBD = Colorblindness, deutan (Xq28) (F)
CBP = Colorblindness, protan (Xq28) (F)
CBS = Cystathionine beta-synthase - chr. 21 (S)
CBT = Carotid body tumor - ?13q34 (F)
CC = Congenital cataract - (?16q; ?linked to HP) (F)
CDPX = Chondrodystrophia punctata, X-linked - ?Xp22.32 (Ch)
CES = Cat eye syndrome - ?22pter-q11 (D)
CG = Chorionic gonadotropin (?nonstructural locus)(?chr. 10 and 18) (S, REb)
CGA = Chorionic gonadotropin, alpha chain - 6q11-q21 (?18p11)(REa)
CGB = Chorionic gonadotropin, beta chain - chr. 19 (REa)
CGD = Chronic granulomatous disease - (Xp22; linked to Xg) (F)
CHE1 = Pseudocholinesterase-1 - (chr. 3; linked to TF) (F)
CHR = Chromate resistance - 5q35 (S)
CKBB = Creatine kinase, brain type - 14q32-qter (?also 17) (S)
CLGN(CLG) = Collagenase (recessive dystrophic epidermolysis bullosa) - chr. 11 (S)
CML = Chronic myeloid leukemia - 22q11.3 (Ch)
CMT1 = *Charcot-Marie-Tooth disease* - (1q; linked to Fy) (F)
Co = Colton blood group (chr. 2; linked to Jk) (D, F)
COI = Coloboma of iris - ?2p25.1-pter (Ch)
COL1A1 = Collagen I alpha-1 chain - 17q21O-q220 (S, M, A, REa)
COL1A2 = Collagen I alpha-2 chain - 7q21-q22 (S, REa, D)
COL3A1 = Collagen III alpha-1 chain - chr. 7 (S)
COL4A1 = Collagen IV alpha-1 chain - ?chr. 17 (REa)

COLM = Collagen, alpha-1(I)-like - chr. 12 (REa)
CP = Ceruloplasmin - (chr. 3; linked to TF) F
CPA = Carboxypeptidase A - 7q22-qter (REa)
CPRO = Coproporphyrinogen oxidase - chr. 9 (S)
CPSD = Cathepsin D - 11pter-q12 (S)
CRP = C-reactive protein - chr. 1 (REa)
CS = Citrate synthase, mitochondrial - 12p11-qter (S)
CSCI = Corticosterone side-chain isomerase - (?6p; ?linked to MHC) (H)
CSA,CSB(CSH1, CSH2) = Chorionic somatomammotropin (see PL) - 17q21O-q22O (S, REa, A)
CSL = Chorionic somatomammotropin-like - 17q21O-q22O (RE)
CTH = Cystathionase - chr. 16 (S)
CTRB = Chymotrypsinogen B - chr. 16 (S or REa)
CVS = Coronavirus 229E sensitivity - 15q11-qter (S)

D1S3 = Previously anonymous DNA segment found identical to SK (q.v.)
D14S1 = DNA segment - 14q32 (S)
DBH = Dopamine-beta-hyroxylase - (?chr. 9; ?linked to ABO) (F)
DC1 = Ia (immune response) antigens of DC1 specificity - 6p2105-p23 (F)
DCE = Desmosterol-to-cholesterol enzyme - chr. 20 (F)
DGI1 = Dentinogenesis imperfecta-1 - (chr. 4; linked to GC) (F)
DGS = DiGeorge syndrome - 22q11 (Ch)
DHFR = Dihydrofolate reductase - 5q11-q22 (S, REa, H)
DHPR(QDPR) = Quinoid dihydropteridine reductase - chr. 4 (S)
DHTR = Dihydrotestosterone receptor = TFM (q.v.)
DIA1 = NADH-diaphorase - 22q1331-qter (S)
DIA2 = Diaphorase-2 - ?chr. 7 (S)
DIA4 = Diaphorase-4 - 16q12-q21 (S)
DIPI(VDI) = Defective interfering particle induction, control of - chr. 16 (S)
DJS = Dubin-Johnson syndrome - chr. 13 (LD)
DLX1 = Dyslexia-1 - chr. 15 (Fc)
DMD = Duchenne muscular dystrophy - ?Xp12-p21 (Ch)
DM = Myotonic dystrophy - (chr. 19; in group linked to C3) (F)
DNCM = Cytoplasmic membrane DNA - 9qh (H, A)
DNL = Lysosomal DNA-ase - chr. 19 (S)
Do = Dombrock blood group - (1p; linked to PGD) (F)
DTS = Diphtheria toxin sensitivity - 5q15-qter (S)

E11S = Echo 11 sensitivity - 19q (S)
EBR1 = CLGN (q.v.)
EBS1 = Epidermolysis bullosa, Ogna type (?chr. 16; linked to GPT1) (F)
EGFR = Epidermal growth factor, receptor for - 7p13-p22 (S)
EL1 = Elliptocytosis-1 - (1p; linked to Rh) (F)
EL2 = Elliptocytosis-2 - (?linkage to chr. 1 markers other than Rh) (F)
ELA1 = Elastase-1 - chr. 12 (REa)
EMTB(RPS14) = Emetine resistance (ribosomal protein S14) - 5q31-q35 (S)
ENO1 = Enolase-1 - 1p36-1pter (S, F, R)
ENO2 = Enolase-2 - 12p11-p12 (S)
ERBA = Oncogene:avian erythroblastic leukemia virus - chr. 17 (S)
ERBB = Oncogene ERBB - 7pter-q22 (S)
ERV1 = Endogenous retrovirus-1 - chr. 18 (S)
ESA4 = Esterase-A4 - 11q13-q22 (S)
ESAT = Esterase activator - chr. 14 (S)
ESB3 = Esterase-B3 - chr. 16 (S)
ESD = Esterase D - 13q14.1 (S, F, D)
EXT = Multiple exostosis - (?9q3; ?linked to ABO) (F)

F7 = Clotting factor VII - 13q34 (D)
F7E = Clotting factor VII expression (chr. 8) (D)
F8C = Clotting factor VIII, procoagulant component = HEMA (q.v.)
F9 = Clotting factor IX = HEMB (q.v.)
F10 = Clotting factor X - 13q34 (D)
F12(HAF) = Clotting factor XII (Hageman factor) - 6p23-pter (D)
F13A = Factor XIII, component A - (6p, linked to MHC) (F)
FCP = F-cell production - (11p)(F)
FDH = Formaldehyde dehydrogenase - 4p14-qter (S)
FEA = F9 embryonic antigen - (?6p; ?linked to HLA) (H)
FES = Onc gene: feline sarcoma virus - 15q25-q26 (S)

FGA = Fibrinogen, alpha chain - 4q21-q31 (REa, H)
FGB = Fibrinogen, beta chain - 4q21-q13 (REa)
FGG = Fibrinogen, gamma chain - (chr. 4; linked to MNSs) (F, REa)
FH = Fumarate hydratase - 1q42.1(S, R, D)
FHC(HC) = Familial hypercholesterolemia - 19pter-q13 F, S
FMS = Oncogene FMS (McDonough feline sarcoma virus) - 5q34 (S)
FN1 = Fibronectin - chr. 2 (S) (see also chr. 8 and 11)
FOS = Oncogene FOS: FBJ osteosarcoma virus - 2q22-q34 (REa, A)
FPGS = Folylpolyglutamate synthetase - 9cen-9q34 (S)
FS = FRAGILE SITE, observed in cultured cells, with or without folate deficient medium, or BrdU - 2q11.2; 2q13; 3p14.2; 6p23; 6q26; 7p11.2; 8q22.3; 9p21.1; 9q31; 10q23.3; 10q25.2; 11q13.3; 11q23.3; 12q13.1; 16p12.3; 16q22.1; 16q23; 17p12; 20p11.23; Xq26; Xq27.3.
FTH = Ferritin heavy chain - chr. 19 (S)
FTL = Ferritin light chain - chr. 19 (S)
FUCA1 = Alpha-L-fucosidase-1 - 1p32-p34 (S, F, R)
FUSA2 = Alpha-L-fucosidase-2 - (?chr. 4; linked to PLG) F
FUSE = Polykaryocytosis inducer - chr. 10 (S)
Fy = Duffy blood group - 1q12-q21 (F, Fc)

GAA = Acid alpha-glucosidase - chr. 17 (S)
GALB = Alpha-galactosidase B - 22q13-qter (S)
GALE = Galactose-4-epimerase - 1p32-pter (S, LD)
GALK = Galactokinase - 17q21-q22 (S, C, R)
GALT = Galactose-1-phosphate uridyltransferase - 9p13-p21 (S, D, F)
GANAB = Neutral alpha-glucosidase AB - 11q13-qter (S)
GANC = Neutral alpha-glucosidase C - chr. 15 (S)
GAPD = Glyceraldehyde-3-phosphate dehydrogenase - 12p13 (S, D)
GARS = Glycinamide ribonucleotide synthetase - chr. 21 (S)
GBA = Acid beta-glucosidase - 1q (S)
GC = Group-specific component - 4q11-q13 (F, Fc)
GCF1 = Growth rate controlling factor-1 - chr. 7 (S)
GCF2 = Growth rate controlling factor-2 - chr. 16 (S)
GCG = Glucagon - 2p36-p37 (REa,A)
GCPS = Greig craniopolysyndactyly syndrome - ?3p21.1 or 7p13 (Ch)
GCTG = Gamma-glutamylcyclotransferase - 7p14-pter (S)
GDH = Glucose dehydrogenase - 1pter-p36.13 (S)
GH-PL = GROWTH HORMONE/PLACENTAL LACTOGEN GENE FAMILY - 17q210-q220 (S, REa, A)
GHN = Growth hormone, normal - 17q210-q220 (S, REa, A)
GHS = Goldenhar syndrome - ?7p (Ch)
GHV = Growth hormone, variant - 17q210-q220 (S, REa, A)
GLA = Alpha-galactosidase A - Xq22-q24 (F, S, R)
GLAT = Galactose + activator - chr. 2 (S)
GLAU1 = Congenital glaucoma-1 - ?chr. 11 (Ch)
GLB1 = Beta-galactosidase-1 - 3p21-cen (S)
GLB2 = Beta-galactosidase-2 - 22q13-qter (S)
GLO1 = Glyoxalase I - 6p21.3-p21.2 (F,S)
GLYB = Glycine auxotroph B, complementation of hamster - 8q21.1-qter (S)
GOT1 = Glutamate oxaloacetate transaminase, soluble - 10q25.3-q26.1 (S, D)
GOT2 = Glutamate oxaloacetate transaminase, mitochondrial - 16p12-q22 (S)
G6PD = Glucose-6-phosphate dehydrogenase - Xq28 (F, S, R)
GPD1 = Alpha-glycerophosphate dehydrogenase (glycerol-3-phosphate dehydrogenase) - chr. 12 (S)
GPHB = GLYCOPEPTIDE HORMONE BETA CLUSTER - chr. 19
GPI = Glucosephosphate isomerase - 19p13-q13 (S, D)
GPT1 = Glutamate pyruvate transaminase, ?soluble red cell - ?16pter-p11 (S)
GPT2 = Glutamate pyruvate transaminase, ?soluble liver - ?8q13-qter (S, EM)
GPX1 = Glutathione peroxidase-1 - 3p13-q12 (S)
GRL = Glucocorticoid receptor, lymphocyte - chr. 5 (S)
GRP78 = Glucose-regulated protein, GRP78 - 9cen-9qter (REa)
GRS = Gardner syndrome - ?2q14.3-q21.3 (Ch)
GSAS = Glutamate-gamma-semialdehyde synthetase - chr. 10 (S)
GSR = Glutathione reductase - 8p21.1 (S, D)
GST3 = Glutathione S-transferase-3 - 11q13-q22 (S)
GUK1 & 2 = Guanylate kinase-1 & 2 - 1q32-q42 (S)
GUSB = Beta-glucuronidase - 7cen-q22 (S)
GUSM = Beta-glucuronidase modifier - chr. 19 (S)

H = HISTONE GENE FAMILY (H1, H2A, H2B, H3, H4) - 7q32-q36 (A)
HADH = Hydroxyacyl-CoA dehydrogenase - chr. 7 (S)
HAF = F12 (q.v.)
HAGH = Glyoxalase II (hydroxyacyl glutathione hydrolase) - chr. 16 (S)
HBA = Hemoglobin alpha chain - 16p12-pter (HS, REa, A, D).
HBAC = AC (q.v.)
HBB = Hemoglobin beta chain - 11p15 (LD, AAS, F, RE, S)
HBBC = NAC (q.v.)
HBD = Hemoglobin delta chain - 11p15 (LD, AAS, F, RE, S)
HBG1 = Hemoglobin gamma chain, ala as AA 136 - 11p15 (AAS, RE)
HBG2 = Hemoglobin gamma chain, gly as AA 136 - 11p15 (AAS, RE)
HBGR = Hemoglobin gamma regulator - 11p15 (RE)
HBHR = Hemoglobin H related mental retardation - ?16p (F)
HBE = Hemoglobin epsilon chain - 11p1205-p1208 (AAS, RE)
HBZ = Hemoglobin zeta chain - 16p12-pter (REa, A)
HBZP = Hemoglobin zeta pseudogene - 16p12-pter (REa, A)
HC = FHC (q.v.)
HCVS = CVS (q.v.)
HD = Huntington disease - chr. 4 (Fd)
HEMA(F8C) = Classic hemophilia (hemophilia A) - (Xq28) (F)
HEMB(F9) = Hemophilia B - (Xq26-q27) REa, A, F
HEXA = Hexosaminidase A - 15q22-152q25.1 (S)
HEXB = Hexosaminidase B - 5q13 (S)
HFE = Hemochromatosis (chr. 6; linked to HLA) (LD,F)
Hh = Bombay phenotype - (chr. 19; linked to Lutheran) (F)
HK1 = Hexokinase-1 - 10pter-p11 (S)
HLA-A,B,C = Human leukocyte antigens - 6p21.3 (F, S, A)
HLA-D/DR = Human leukocyte antigen, D-related - 6p21.3 (F, S, A)
HLA-DC = Human leukocyte antigen, DC type - 6p21.3 (F, A)
HLA-SB = Human leukocyte antigen, SB type - 6p21.3 (F, S, A)
HP = Haptoglobin - 16q22 (Fc)
HPA1 = Hpa I restriction endonuclease polymorphism - 11p1205-p1208 (RE)
HPRT = Hypoxanthine-guanine phosphoribosyltransferase - Xq26-q27 (F, S, R)
HRAS1(RASH1) = Harvey rat sarcoma-1 protooncogene - 11p15.5-p15.1 (S)
HRAS2(RASH2) = Harvey rat sarcoma-2 protooncogene - X chr. (REa) - pseudogene
HTOR = 5-Hydroxytryptamine oxygenase regulator - chr. 21 (D)
HV1S = Herpes virus-sensitivity (?chr. 3 and/or 11) (S)
HYA = Y histocompatibility antigen, locus A - Y chr. (F)
HYB = Y histocompatibility antigen, locus B - X chr. (Ch) - ?regulator
HYC = Y histocompatibility antigen, locus C - ?X chr. (Ch) - ?receptor

IDA(IDUA) = Alpha-L-iduronidase - 22pter-q11 (S)
IDDM = Insulin-dependent diabetes mellitus - (?chr.6; ?linked to HLA) (F)
IDH1 = Isocitrate dehydrogenase, soluble - 2q32-qter (S)
IDH2 = Isocitrate dehydrogenase, mitochondrial - 15q21-qter (S)
IF1, IF2 = Interferon 1 and 2 - ?chr. 2 and 5, respectively (S)
IFF(IFB) = Interferon, fibroblast - 9p24-p13 (REa)
IFI(IFG) = Interferon, immune - 12q24.1 (A)
IFL(IFA) = INTERFERON, LEUKOCYTE, GENE CLUSTER - 9qter-p13 (REa)
IFR = AVR (q.v.)
IFRC = Interferon receptor (antiviral protein) - 21q21-qter (S, D)
IGAS = Immunoglobulin heavy chains attachment site - chr. 2 (S)
IGEP = IgE pseudogene - chr. 9 (A)
IGH = IMMUNOGLOBULIN HEAVY CHAIN GENE CLUSTER - 14q32.3 (S)
IGHA1 = Gene for constant region of heavy chain of IgA1 - 14q32 (S, RE)
IGHA2 = Gene for constant region of heavy chain of IgA2 - 14q32 (S, RE)
IGHD = Gene for constant region of heavy chain of IgD - 14q32 (S, RE)
IGHE = Gene for constant region of heavy chain of IgE - 14q32 (S, RE)
IGHG1 = Gene for constant region of heavy chain of IgG1 - 14q32 (S, RE)
IGHG2 = Gene for constant region of heavy chain of IgG2 - 14q32 (S, RE)
IGHG3 = Gene for constant region of heavy chain of IgG3 - 14q32 (S, RE)
IGHG4 = Gene for constant region of heavy chain of IgG4 - 14q32 (S, RE)
IGHJ = Gene (multiple) for J (joining) region of heavy chain - chr. 14 (S, RE)
IGHM = Gene for constant region of heavy chain of IgM - chr. 14 (S, RE)
IGHV = Gene (multiple) for variable region of heavy chain - chr. 14 (S, RE)
IGK = IMMUNOGLOBULIN KAPPA LIGHT CHAIN GENE CLUSTER - 2p (A)

IGKC = Gene for constant region of kappa light chain - 2p (A)
IGKJ = Gene (multiple) for J (joining) region of kappa light chain - 2p (A)
IGKV = Gene (multiple) for variable region of kappa light chain - 2p (A)
IGL = IMMUNOGLOBULIN LAMBDA LIGHT CHAIN GENE CLUSTER - chr. 22 (REa)
IGLC = Gene for constant region of lambda light chain - chr. 22 (REa, A)
IGLJ = Gene (multiple) for J (joining) region of lambda light chain - chr. 22 (REa)
IGLV = Gene (multiple) for variable region of lambda light chain - chr. 22 (REa)
IHC = Immune response to synthetic polypeptide - HGAL - 6p21.3 (F)
IL2 = TCGF (q.v.)
INS = Insulin - 11p15-p15.5 (S,A,REb)
INSL = Insulin-like DNA sequence - 6p23-q12 (REa)
INT1 = Oncogene INT: putative murine mammary cancer oncogene - 12pter-q14 (REa)
IRDN = Insulin-related DNA polymorphism - 11p13-p15.5 (RE)
IR = BR1A, BR1B, etc. (q.v.) ?homologs of Ir gene in mouse
IS = Immune suppression - 6p2105-6p2300 (F)
ITG(BR1A, BR1B) = Immune response to synthetic polypeptide - TGAL - 6p21.3 (F)
ITPA = Inosine triphosphatase - 20p (S)

Jk = Kidd blood group - (chr. 2; linked to IGKC) (F)

KAR = Aromatic alpha-keto acid reductase - 12p (S)
KRAS1(RASK1) = Kirsten rat sarcoma protooncogene-1 - 6p23-q12 (S) - ?pseudogene
KRAS2(RASK2) = Kirsten rat sarcoma protooncogene-2 - chr. 12 (S)

LAG5 = Leukocyte antigen group five - chr. 4 (S)
LAP = Laryngeal adductor paralysis - (?chr. 6; ?linked to HLA) (F)
LARS(RNTLS) = Leucyl-tRNA synthetase - chr. 5 (S)
LCAT = Lecithin-cholesterol acyltransferase - (16q22; linked to HP)(F, LD)
LCH = Lentil agglutinin binding - chr. 14 (S)
LDHA = Lactate dehydrogenase A - 11p1203-p1208 (S)
LDHB = Lactate dehydrogenase B - 12p121-p122 (S, D)
LDHC = Lactate dehydrogenase C - (?12p; linked to LDHB in pigeon) (H)
Le = Lewis blood group (chr. 19; linked to C3) (F)
LEU7 = Leu-7 membrane antigen of natural killer lymphocytes - chr. 11 (S)
LGS = Langer-Giedion syndrome - 8q23.3 (Ch)
LHB = Luteinizing hormone beta subunit - chr. 19 (RE)
LIPA = Lysosomal acid lipase-A - chr. 10 (S)
LIPB = Lysosomal acid lipase-B - chr. 16 (S)
LSD = Letterer - Siwe disease - ?13q14-q31 (Ch)
Lu = Lutheran blood group - chr. 19 (F)

M130 = External membrane protein-130 - chr. 10 (S)
M195 = External membrane protein-195 - chr. 14 (S)
M4F2 = Monoclonal antibody 4F2, antigen defined by - 11q (S)
M7VS1 = Baboon M7 virus sensitivity-1 - chr. 19 (S)
MA3D8 = Monoclonal antibody A3D8 antigen defined by 11p (S)
MANA = Cytoplasmic alpha-D-mannosidase - 15q11-qter (S)
MANB = Lysosomal alpha-D-mannosidase - 19pter-q13 (S)
MAP97(MFJ1) = Melanoma-associated antigen p97 - chr. 3 (S)
MAR = Macrocytic anemia, refractory - 5q (Ch)
MARS = MTRNS (q.v.)
MDH1 = Malate dehydrogenase, soluble - 2p23 (S)
MDH2 = Malate dehydrogenase, mitochondrial - 7p22-q22 (S)
MDI = Manic-depressive illness (?chr. 6; ?linked to HLA) (F)
MDLS = Miller-Dieker lissencephaly syndrome - ?17p13 (Ch)
ME1 = Malic enzyme, soluble - 6q12 (S)
ME2 = Malic enzyme, mitochondrial - ?chr. 11 (H)
MEN2 = Multiple endocrine neoplasia, type II (Sipple syndrome) - 20p12.2 (Ch)
MHC = MAJOR HISTOCOMPATIBILITY COMPLEX - 6p2105-p23 (F, S)
MLRW = Mixed lymphocyte reaction, weak (chr. 6) (F)
MMC = Malignant melanoma, cutaneous - (?1p; ?linked to Rh) (F)
MN = MN blood group - 4q28-q31 (F, Fc, AAS)
MOS = Onc gene: Moloney murine sarcoma virus - 8q22 (S)
MPI = Mannosephosphate isomerase - 15q22-qter (S)
MRBC = Monkey red blood cell receptor - chr. 6 (S)
MTR = 5-Methyltetrahydrofolate: L-homocysteine S-methyltransferase (tetrahydropteroyl-glutamate methyltransferase) - chr. 1 (S)

MTRNS (MARS) = Methionyl-tRNA synthetase - chr. 12 (S)
MYB = Onc gene: avian myeloblastosis virus - 6p21-q22 (S)
MYC = Onc gene: myelocytomatosis virus - 8q24 (A)
MYHS = MYOSIN, SKELETAL, HEAVY CHAIN GENE FAMILY - 17p11-pter (REa)
MYHSA1 = Myosin, skeletal, heavy chain, adult-1 - 17p11-pter (REa)
MYHSA2 = Myosin, skeleta, heavy chain, adult-2 - 17p11-pter (REa)
MYHSE1 = Myosin, skeletal heavy chain, embryonic-1 - 17p11-pter (REa)

NAG(HBBC) = NON-ALPHA GLOBIN CLUSTER (HEMOGLOBIN BETA CLUSTER) - 11p1205-1208 (S, RE)
NAGA = N-acetyl-alpha-D-galactosaminidase - 22q13 (S)
NARS = ASNRS (q.v.)
NB = Neuroblastoma - 1p32-1pter (?1p34) (Ch)
NDF = Neutrophil differentiation factor (chr. 6) (LD)
NEU1 = Neuraminidase-1 - ?6p2105-6p2300 (?linked to HLA) (H)
NGF = Nerve growth factor - 1p21-1pter (REa)
NHCP1 = Nonhistone chromosomal protein-1 - chr. 7 (S)
NHCP2 = Nonhistone chromosomal protein-2 - chr. 16 (S)
NF1 = Neurofibromatosis - (?chr. 19; linked to DM) (F)
NF2 = Neurofibromatosis - (?chr. 4; linked to GC) (F)
NF3 = Familial intestinal neurofibromatosis - ?12q21-q24.2 (Ch)
NLP1 = Neoplastic lymphoproliferation-1 - ?8q24.3 (Ch)
NLP2 = Neoplastic lymphoproliferation-2 - ?18q24 (Ch)
NM = Neutrophil migration - 7q22-qter (D)
NP = Nucleoside phosphorylase - 14q13 (S, D)
NPS1 = Nail-patella syndrome - (9q3; linked to AK-1) (F)
NRAS = Oncogene NRAS - 1p (REa)

OA1 = Ocular albinism-1 (Nettleship-Falls type) - (Xp22; linked to Xg) (F)
OA2 = Ocular albinism-2 (Forsius-Eriksson type) - (Xp22; linked to Xg) (F)
OAK = Optic atrophy, Kjer type - (?chr. 2; ?linked to Jk) (F)
OIAS = 2',5'-oligoisoadenylate synthetase - chr. 11 (S)
OMPD = Orotidylmonophosphate decarboxylase (with OPRT, q.v.)
OMR(ATPM) = Oligomycin resistance (mitochondrial ATPase) - chr. 10 (S)
OPRT = Orotate phosphoribosyltransferase - OMP decarboxylase - 3cen-q21 (S)
ORM = Orosomucoid - (9q34; linked to ABO) (F)
OTC = Ornithine transcarbamoylase - Xp21 (Ch)

PAH(PKU) = Phenylalanine hydroxylase - chr. 12 (REa)
PAIS = Phosphoribosylaminoimidazole synthetase - chr. 21 (S)
PBGD(UPS) = Porphobilinogen deaminase - 11q23-qter (S)
PDB = Paget disease of bone - (?chr. 6; ?linked to HLA) (F)
PDGF = Platelet derived growth factor = SIS (q.v.)
PEPA = Peptidase A - 18q23 (S, D)
PEPB = Peptidase B - 12q21 (S)
PEPC = Peptidase C - 1q (S, R)
PEPD = Peptidase D - 19pter-q13 (S)
PEPS = Peptidase S - 4p12-q12 (S)
PFGS = Phosphoribosyl formylglycinamidine synthetase - chr. 14 (S)
PFKL = Phosphofructokinase, liver - 21q22 (S)
PFKM = Phosphofructokinase, muscle - 1cen-1q32 (S)
PFKP(PFKF) = Phosphofructokinase, platelet - 10pter-p11.1 (S)
PG = Pepsinogen - ?chr. 6 (F,H)
PGAM1 = Phosphoglycerate mutase A - 25.3-q26.1 (D)
PGD = 6-Phosphogluconate dehydrogenase - 1p36.13-pter (F-S)
PGFT = Phosphoribosylglycinamide formyltransferase - 14q22-qter (S)
PGK = Phosphoglycerate kinase - Xq13 (F, S)
PGM1 = Phosphoglucomutase-1 - 1p221 (F, S, R)
PGM2 = Phosphoglucomutase-2 - 4p14-q12 (S)
PGM3 = Phosphoglucomutase-3 - 6q12 (S, F, OT)
PGP = Phosphoglycolate phosphatase - 16p13-p12 (S)
PI = Alpha-1-antitrypsin - 14q (F,S)
PKM2 = Pyruvate kinase-3 - 15q22-qter (S,D)
PKU = PAH (q.v.)
PL = Placental lactogen (same as CSA, CSB, q.v.) - 17q210-17q220 (S, REa, A)
PLA = Plasminogen activator - chr. 6 (S)
PLG = Plasminogen - chr. 4 (S)
PLT1 = Primed lymphocyte test-1 - 6p2105-p23 (F)
POMC = Proopiomelanocortin - 2p23 (REa)

PP = Inorganic pyrophosphatase - 10q11.1-q24 (S)
PPAT = Phosphoribosylpyrophosphate amidotransferase - 4pter-q21 (S)
PRGS = Phosphoribosylglycinamide synthetase - 21q22 (S,H)
PRL = Prolactin - 6p23-q12 (S)
PRPS = Phosphoribosylpyrophosphate synthetase - X chr. (F, S)
PSP = Phosphoserine phosphatase - 7qter-q22 (S)
PTH = Parathyroid hormone - 11pter-p11 (REa)
PVS = Polio virus sensitivity - 19q (S)
PWS = Prader-Willi syndrome - 15q11-q12 (Ch)

1qh = Centric heterochromatic segment, long arm, chr. 1 (formerly "uncoiler")
9qh = Centric heterochromatic segment, long arm, chr. 9
QDPR = DHPR (q.v.)

RACH = Regulator of acetylcholinesterase - chr. 2 (D)
RAF1 = Oncogene RAF1 - chr. 3 (S)
RAF2 = Oncogene RAF2 - chr. 4 (S)
RB1 = Retinoblastoma-1 - 13q14.1 (Ch)
RCC = Renal cell carcinoma - ?3p21 (Ch)
Rd = Radin blood group - (1p32-p34; linked to Rh) F
REN = Renin - 1p21-qter (REa)
Rh = Rhesus blood group (1p32-p36.11) (F-S,D)
RGS = Rieger syndrome - ?4q23-q27 (Ch)
RN5S = 5S RNA gene(s) - 1q42-q43 (A)
RNTMI(TRM1,2) = Initiator methionine tRNA - 6p23-q12 (REa)
RNR = RIBOSOMAL RNA - 13p12, 14p12, 15p12, 21p12, 22p12 (A)
RNU1 = RNA, U1 small nuclear - 1p36 (S)
RP1 = Retinitis pigmentosa-1 (?chr. 1) (F)
RPE = Ribulose 5-phosphate 3-epimerase - 2q32-qter (S)
RPS14 = Ribosomal protein S14 = EMTB (q.v.)
RS = Retinoschisis - (Xp22; linked to Xg) (F)
RWS = Ragweed sensitivity - (chr. 6; ?linked to HLA) (F)

SAHH(AHCY) = S-adenosylhomocysteine hydrolase - 20 cen-20q13.1(S)
Sc = Scianna blood group - (1p32-p34) (F)
SCA1 = Spinocerebellar ataxia I - (chr. 6; linked to HLA) (F)
SCCL = Small-cell cancer lung - 3p14-p23 (Ch)
SDH = Succinate dehydrogenase - 1p22.1-qter (S)
Se = Secretor - (chr. 19; in group linked to C3) F
Sf = Stoltzfus blood group - (4q; linked to MNSs) (F)
SGP75 = Surface antigen, glycoprotein 75,000 - chr. 11 (S)
SHMT = Serine hydroxymethyltransferase - 12q12-q14 (S)
SIS(PDGF) = Oncogene: simian sarcoma virus - chr. 22 (S)
SK(D1S3) = Oncogene Sloan-Kettering chicken virus - 1q12-1qter (REa)
SOD1 = Superoxide dismutase, soluble - 21q21 (S,D)
SOD2 = Superoxide dismutase, mitochondrial - 6q21 (S)
SORD = Sorbitol dehydrogenase - 15pter-q21 (S, H)
SP3 = Spermatogenesis factor-3 (azoospermia third factor) - ?Yq11 (D)
SPA2, SPA5 = Surface polypeptide, anonymous - chr. 2, 5, respectively (S)
SPC = SALIVARY PROTEIN COMPLEX - ?6p (F)
SPH = Spherocytosis - 8p11, ?14q (F, Ch)
SRC = Oncogene SRC (Rous sarcoma) - chr. 20 (REa)
Ss = Ss blood group - 4q28-q31 (F, Fc, AAS)
SST = Somatostatin - 3q27-3q28 (REa, A)
STA = Stature - Y chr. (D)
STS = Steroid sulfatase - Xp22.3 (F, S)

TCGF(IL2) = T-cell growth factor (interleukin-2) - chr. 4 (REa)
TCN2 = Transcobalamin II - ?chr. 16 or 17 (H)
TDF = Testis determining factor - Y chr. (?same as HYA) (F)
TF = Transferrin - chr. 3 (S, H)
TFM(DHTR) = Testicular feminization syndrome (dihydrotestosterone receptor)- Xp11-q13 (S)
TFR = Transferrin receptor - chr. 3 (S, H, REa)
TG = Thyroglobulin - chr. 8 (S)
THC = Thrombocytosis, primary - 21q (Ch)
TK1 = Thymidine kinase, soluble - 17q21-q22 (S, C ,R)
TK2 = Thymidine kinase, mitochondrial - chr. 16 (S)

TKCR = TKCR syndrome of Goeminne - ?Xq28 (Ch)
TPI1 & 2 = Triosephosphate isomerase-1 & 2 - TPI-1 on 12p13 (S, D)
TRC = T-cell receptor for MHC antigens - ?chr. 14 (H)
TRM1,2 = RNTMI (q.v.)
TRY1 = Trypsin-1 - 7q22-qter (REa)
TS = Tooth size - ?Yq11 (D)
TYS = Sclerotylosis - (4q; linked to MNSs) (F)

UGP1 = Uridyl diphosphate glucose pyrophosphorylase-1 - 1q21-q23 (S, R)
UGP2 = Uridyl diphosphate glucose pyrophosphorylase-2 - chr. 2 (S)
UMPK = Uridine monophosphate kinase - 1p32 (S, R)
UP = Uridine phosphorylase - chr. 7 (S)
UPS = Uroporphyrinogen I synthase = PBGD (q.v.)
UVDR = Ultra-violet damage repair - chr. 13 (S)

VDI = DIPI (q.v.)
VMD1 = Macular dystrophy, atypical vitelliform - ?16p (F)

WAGR = Wilms tumor/aniridia/gonadoblastoma/retardation - 11p13 (Ch)
WARS = Tryptophanyl-tRNA synthetase - 14q21-qter (S)
WS1 = Waardenburg syndrome-1 - (?9q34; ?linked to ABO) (F)

Xg = Xg blood group (Xp22.3; linked to STS) (F)

Xk = Kell blood group precursor (Xp22.3; linked to Xg) (F)
XPA = Xeroderma pigmentosum A complementation - 1q (S)
XRS = X-ray sensitivity - ?13q14 (Ch)

Figure 2

Four Commingling Methodologic Streams in Gene Mapping

	Family Studies	Chromosome Studies	Somatic Cell Studies	Molecular Studies
F	Linkage (F) (Linkage disequilibrium, LD)	Linkage with heteromorphism or rearrangement (Fc) Deletion mapping (D)	(Homology of synteny,H)	Linkage with RFLPs (Fd)
CH		Dosage effect (D) Exclusion mapping (EM) Chromosome aberration (Ch) Virus-induced changes (V) Fragile sites (FS)	Assignment by SCH (S) Regional mapping by SCH (S) Chromosome-mediated gene transfer, CMGT	In situ hybridization (A) Molecular analysis of flow sorted chromosomes (REb)
Methods of Autosomal Mapping		S	SCH synteny test (S) Radiation induced gene segregation (R) Microcell-mediated gene transfer, MCGT (M)	DNA or RNA hybridization in solution (HS) Restriction enzyme analysis (REa) DNA-mediated gene transfer, DMGT (DM)
			M	Restriction enzyme fine mapping (RE) DNA sequencing (NA) AA sequencing (Lepore approach)(AAS)

I. GENE MAP OF THE AUTOSOMES

About 1600 loci are known with confidence to exist on autosomes, on the basis mainly of characteristic patterns of inheritance of alternative forms of particular traits (see Table II). As indicated by the following, some mapping information is available concerning over 30% of these loci. The regional assignments on the left represent SROs (shortest regions of overlap) of various assignments using broken or rearranged chromosome). The regional assignments in parentheses include information concerning gene order to arrive at a shorter region of assignment, or represent assignments made indirectly through linkage with an assigned locus. In addition to the loci listed here, anonymous DNA segments, antigens defined by monoclonal antibodies, surface antigens, and function-unknown protein spots have been assigned to individual autosomes, as catalogued by HGM 7.

A. Chromosomal assignments (see Fig. 1)

Chromosome No. 1

1p36	RNA, U1 small nuclear (18068) REa, A
distal to 1p36.13	(P)Adenovirus-12 chromosome modification sites-1A (10292) V
1p(distal to Rh?)	(L)Retinitis pigmentosa-1 (18010) F
?1p(?linked to Rh)	(L)Malignant melanoma, cutaneous (15560) F (lod2.0, theta 0.30)
1p (12cM distal to PGD?)	(P)Dombrock blood group (11060) F (linked to PGD only in female)
1p36.13-1pter	Glucose dehydrogenase (13809) S, F
1p36.13-1pter	Enolase-1 (17243) S, F, R
1p36.13-1pter	6-Phosphogluconate dehydrogenase (17220) F, S
1p32-1pter (?1p34)	(P)Neuroblastoma (25670) Ch
(1p32-1pter)	Elliptocytosis-1 (13050) F
1p32-1p36 (?1p36.11)*	Rhesus blood group (11170) FS, D
1p32-1pter	UDP galactose-4-epimerase (23035) S, LD
?1p32	(P)Oncogene BLYM: chicken bursal lymphoma (16483) REa
1p34	Alpha-L-fucosidase (23000) S, F, R
(1p32-1p34)	Scianna blood group (11175) F
(1p32-1p34)	(P)Radin blood group (11162) F - ?same as Scianna
cen-PGM1-@8cM-C8-UMPK	(P)C8 (beta polypeptide) (12096) F — not closely linked,
	(P)C8 (alpha-gamma polypeptide) (12095) F — not closely linked,
1p32 (distal to PGM1)	Uridine monophosphate kinase (19171) S, R
1p34	Adenylate kinase-2, mitochondrial (10302) S, F, R
1p22.1	Phosphoglucomutase-1 (17190) F, S, R
1pter-1p23	(P)Alcohol dehydrogenase-2 (beta polypeptide) (10372) S
1p21-1pter	(P)Nerve growth factor (16203) REa
1p21-1qter	(P)Renin (17982) REa
1p13 or 1q25	(L)Aspermiogenesis factor (10845) Ch
1p21	Amylase, pancreatic (10465) F, A, REa — Multiple genes
1p21	Amylase, salivary (10470) F, A, REa — Multiple genes
1p22.1-1p31.1;1p13;1p11-1p21	Oncogene NRAS (16479) REa (?same as NGF)
1qh	Satellite DNA III (D1Z1) (12634) A
(1q2)	(L)Elliptocytosis-2 (13060) F
1q (close to Fy)	Cataract, zonular pulverulent (11620) F
1q12-1qter	(P)Oncogene Sloan-Kettering, SK, chicken virus (16478) REa (=D1S3)
1p21-1qter	Actin, skeletal muscle alpha chains (10261) REa
(I)1q42; 1q21-1q31	Acid beta-glucosidase (glucocerebrosidase) (23080)S
Prox. 1p or 1q	(P)C-reactive protein (12326) REa
1q12-q21 (distal to 1qh)	Duffy blood group (11070) F, Fc
(about 15 cM)	Charcot-Marie-Tooth disease, slow nerve conduction type (11820) F
1q21-1q23	Uridyl diphosphate glucose pyrophosphorylase-1 (19175) S, R
1q22	(P)Adenovirus-12 chromosome modification site-1B (10294) V
(I)1q25;1q42	Peptidase C (17000) S, R
1q42-1q43	5S ribosomal RNA gene(s) (18042) A
1q42.1	Fumarate hydratase (13685) S, R, D
1q32.1-1q42	Guanylate kinase-1 (13927) S,D — Genetic independence
	Guanylate kinase-2 (13928) S,D — unproved
1q42 and 1q22	(P)Adenovirus-12 chromosome modification site-1B and 1C (10293) V
1q23.1-1q23.9(@10cM from Fy)	Antithrombin III (10730) F, D, A, REa
	(P)5-Methyltetrahydrofolate: L-homocysteine S-methyltransferase (tetrahydropteroyl-glutamate methyltransferase) (15657) S
1p22.1-qter	(P)Succinate dehydrogenase (1 of 2 polypeptides) (18547) S
1cen-1q32	(P)Phosphofructokinase, muscle type (23280) S
1q	(P)Xeroderma pigmentosum A (27870) S

In addition: 2 anonymous DNA segments, 5 O'Farrell protein spots, and 2 antigens defined by monoclonal antibodies (HGM 7).

The order of closely linked loci (ENO1 and 6PGD; of Ell, Rh, and FucA; of UMPK and Sc; and of Fy and Cae) is uncertain. "However, the following order of loosely linked segments seems established": 6PGD, Rh, UMPK, PGM1, Amy, lqhl2, Fy, Pep C. (From Rao et al., Am. J. Hum. Genet. 31: 680-696, 1979.)

*Location surmised from genetic distances considered in relation to the chiasma map of lp (see Fig. 3B).

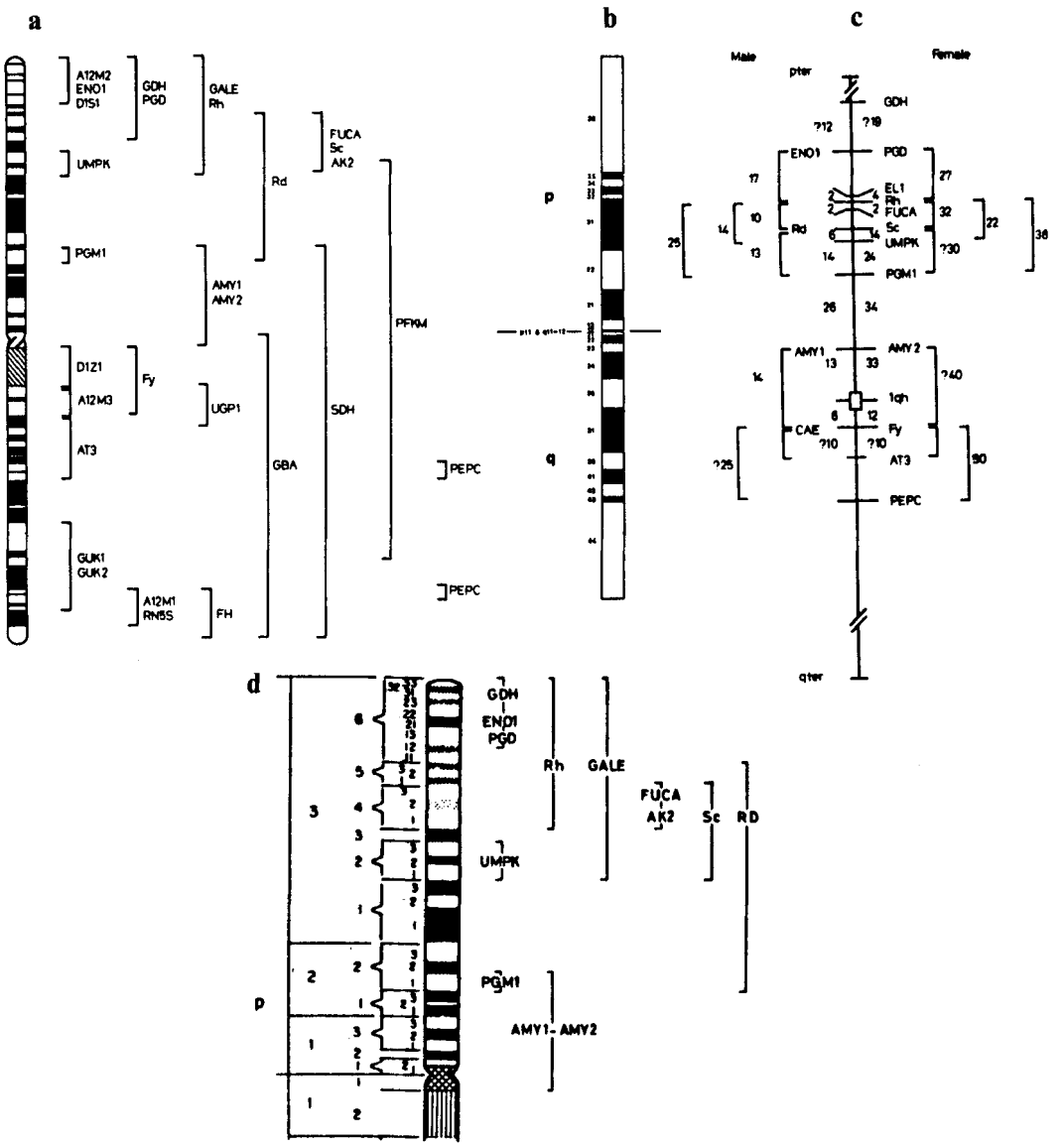


Fig. 3. Maps of chromosome 1 derived from physical assignments (a), observed chiasmata (b), and family linkage studies (c). From Cook and Hamerton, HGM6, Cytogenet. Cell Genet. 32:117, 1982. (Possibly due to lqh, the genetic length of lq is slightly less than that of lp.) (d) Revised order and SROs for lp (Carritt et al., Ann. Hum. Genet. 46:330,1982).

Chromosome No. 2

2p	Aryl hydrocarbon hydroxylase (10833) S
(2p23)	(L)Aniridia (10620) F
2p25.1-2pter	(L)Coloboma of iris (12020) Ch
(2p)	(L)Acrokeratoelastoidosis (10185) F (?linked to ACP1, Jk, IGKC)
(I)2p25;2p23	Acid phosphatase-1 (17150) D, S
?2p23 (close to ACP1?)	Proopiomelanocortin (17683) REa
2p23	Malate dehydrogenase, soluble (15420) S
2p11-2p22	(P)Galactose enzyme activator (13703) S
	(P)Immunoglobulin heavy chain attachment site (14710) S
	(P)Acetylcholinesterase regulator, or derepressor (10065) D
2p23-2q32?	Adenosine deaminase complexing protein-2 (10272) S
	(P)Uridyl diphosphate glucose pyrophosphorylase-2 (19176) S
2p12	IMMUNOGLOBULIN KAPPA LIGHT CHAIN GENE FAMILY (Km; Inv) A, REa
about 23 cM	Variable region of kappa light chain (many genes) (14698)
	J region of kappa light chain (several genes) (14697)
	Constant region of kappa light chain (14720)
	(P)Kidd blood group (11100) F
?linked to Jk (lod 2.15, theta 0.14 male, 0.27 female)	(L)Optic atrophy, Kjer type (16540) F
linked to Jk (lod 3.8 at theta 0.29)	(L)Colton blood group (11045) F (chr. 7 suggested by dosage effect in monosomy 7, Hum. Genet. 62:40, 1982)
2q11.2	Fragile site 2q11.2, folic acid type (13661)
2q13	Fragile site 2q13, folic acid type
2q14.3-2q21.3	(L)Gardner syndrome (17530) Ch
2q22-2q34	(P)Oncogene FOS: FBJ osteosarcoma virus (16481) REa, A
2q32-2qter	Ribulose 5-phosphate 3-epimerase (18048) S
2q32-2qter	Isocitrate dehydrogenase, soluble (14770) S
	Fibronectin-1 (13560) S (prob. structural gene; see chr. 8, 11)
2p23-qter	(L)Interferon-1 (?function - in relation to fibroblast FN; not structural) (see 14757) S
2q36-2q37	Glucagon (13803) REa, A

In addition: 2 anonymous DNA segments (HGM 7) and 1 surface polypeptide (18561).

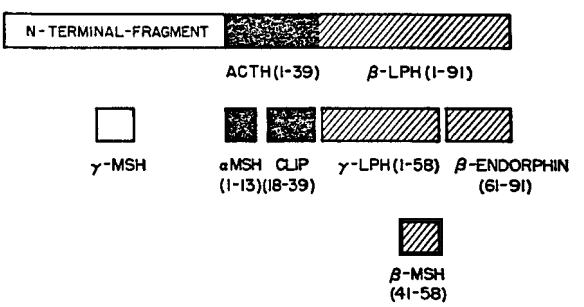


Fig. 4. POMC is the precursor for 7 peptide hormones as indicated here.

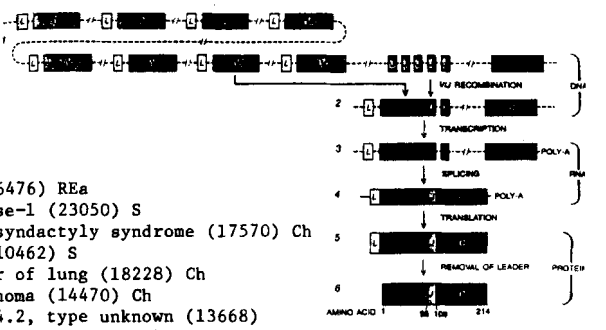


Figure 5. (From Leder, P.: Scientific American 246 (May): 102, 1982.

Chromosome No. 3

	(P)Oncogene RAF1 (16476) REa
3p21-3cen	Beta-galactosidase-1 (23050) S
?3p21.1(or 7p13)	(L)Greig craniopolysyndactyly syndrome (17570) Ch
3p21	Aminoacylase-1 (10462) S
3p14-3p23	(P)Small-cell cancer of lung (18228) Ch
?3p21	(L)Renal cell carcinoma (14470) Ch
3p14.2	Fragile site 3p14.2, type unknown (13668)
3p13-3q12	Glutathione peroxidase-1 (23170) S
3cen-3q21	(P)Orotate phosphoribosyltransferase - OMP decarboxylase (25892) (S)
3q20	Somatostatin (18245) REa
	(P)Temperature sensitive (tsAF8) complement (11695)S
	(P,I)Herpes virus sensitivity (14245) S (see chr. 11)
	(P)Melanoma-associated antigen p97 (15575) S
3q23-3qter	Transferrin receptor (19001) S, H, REa ——?related or identical
	(P)Ceruloplasmin (11770) F, H
	(P)Transferrin (19000) S, H, REa
	(P)Pseudocholinesterase-1 (17740) F——(m, F)*
	(P)Alpha-2HS-glycoprotein (13868) F**

In addition: 4 anonymous DNA segments (HGM 7)
*See Section I, B for definition.
**Probably order = cen-TF-CHE1-A2HS

Chromosome No. 4

@15cM	(L)Alpha-fucosidase-2 (13682) F (linked to PLG; lod 3.63, male theta 0.14)
	(L)Plasminogen (17335) F (?0.28 cM from GC; far from MN)
4pl4-4qter	(P)Formaldehyde dehydrogenase (13649) S
(4q; @25cM from MNSs)	Stoltzfus blood group (11180) F
(4q; linked to MNSs)	Sclerotylosis (18160) F
@11cM	Dentinogenesis imperfecta-1 (12549) F
4q11-4q13	Group-specific component (13920) F, Fc, H, D
?linked to GC	(L)Von Recklinghausen neurofibromatosis (16220) F
4q28-4q31 (by EM)	MN blood group (11130) F, Fc, AAS (male lod 3.79, theta 0.32, GC)
	Ss blood group (11174) F, Fc, AAS
(Linked to MN)	(P)Anterior segment mesenchymal dysgenesis (10725) F
4q23-4q27	(L)Rieger syndrome (18050) Ch
4p12-4q13	Phosphoglucomutase-2 (17200) S
4q11-4q13	Albumin (10360) F (linked to GC), A, REa
4q11-4q13	Alpha-fetoprotein (10415) H, A
4q21-4q31	Fibrinogen, alpha chain (13482) RE, REa, H
4q21-4q31	Fibrinogen, beta chain (13483) RE, REa
(Linked to MNSs)	Fibrinogen, gamma chain (13485) F, REa, H, RE
4p12-4q12	Peptidase S (17025) S, EM
4pter-4q21	(P)Phosphoribosylpyrophosphate amidotransferase (17245) S
?4p (remote from MN, GC)	Huntington disease (14310) Fd
	(P)Quinoid dihydropteridine reductase (26163) S
	(P)Leukocyte antigen group five (LAG5) (15145) S
	(P)Alcohol dehydrogenase, class 1 (10370) REa
	(P)T-cell growth factor (interleukin-2) (18693) REa
	(P)Oncogene RAF2 (16476) REa - pseudogene

In addition: 3 anonymous DNA segments and 1 antigen defined by a monoclonal (HGM 7).
Possible order: PLG - CEN - GC - DGI - Ss - MN - FGG

Chromosome No. 5

5pter-5q1	Leucyl-tRNA synthetase (15135) S, H
5p	(L)Antiviral state repressor regulator (10747) (S)
5q13	Hexosaminidase B (14265) S, Ch
5q15-5qter	Diphtheria toxin sensitivity (12615) S
	Arylsulfatase B (25320) S
5q34	Oncogene FMS (McDonough feline sarcoma) (16477) REa
5q35	Chromate resistance (sulfate transport) (11884) S

5q31-5q35 Emetine resistance (ribosomal protein S14) (13062) S
5q (L)Macrocytic anemia, refractory (15355) Ch
(P)Beta-adrenergic receptor (10969) S
(P)Glucocorticoid receptor, lymphocyte (23157) S
5q11-5q22 Dihydrofolate reductase (12606) S, REa, H (to other chrs. with amplification)
5p (L)Interferon-2 (?function in relation to fibroblast FN; not structural)
(see 14757) S

In addition: 3 anonymous DNA segments (HGM 7)
and 1 surface polypeptide (18561).

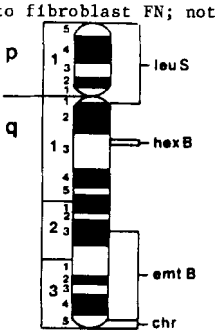


Fig. 6. From Dana and Wasmuth, Molec. Cell Biol. 2:1226, 1982. Ideogram of human chromosome 5 showing provisional regional assignments for the leuS, hexB, emtB and chr genes. (Order on 5q: centromere-LARS-HEXB-EMTB-FMS-CHR-ter Groffen et al., HGM7, 1983.)

Chromosome No. 6

6pter-6p23 (P)Clotting factor XII (Hageman factor) (23400) D
Linked to MHC (P)Clotting factor XIII, A component (13457) F
6p21.3 MAJOR HISTOCOMPATIBILITY COMPLEX (MHC)F, S, A
HLA-A (14280) F About 6cM from centromere
6p21.3 (A) HLA-B (14283) F in one estimate, 14 cM in
order: cen.-D-B-C-A*** HLA-C (14284) F another
HLA-D (14285) F ------ Separateness still HLA-DR alpha
6p21.1 (A) (P)HLA-DR (14286) F------to be established HLA-DR beta
On B side of D (P)HLA-DR2 (14287) F
No crossover with BF Complement component-2 (21700) F,LD - 2% recomb. with HLA-B
Complement component-4F, or C4B (12082) F, H
Complement component-4S, or C4A (12081)F, H
(L)Complement component-4 binding protein (12083) H
(L)Neuraminidase-1 (16205) H
No crossover with C2 Properdin factor B (Bf) (13847) F
Linked to C2, C4, BF Congenital adrenal hyperplasia due to 21-hydroxylase deficiency (20191) F
Near HLA-A end (P)Mixed lymphocyte reaction, weak (15786) F
Centromeric to DR HLA-SB ("secondary B cell response") (14288) F, Ch
Very close to HLA-D/DR HLA-DC (14688) F, A - 2 beta genes
Near HLA-D/DR Primed lymphocyte test-1 (17668) F
(L)Ragweed sensitivity (17945) F
In A/B segment (P)Blastogenic response to synthetic polypeptide - HGAL (14695) F (complementing loci BR2A, BR2B)
(P)Blastogenic response to synthetic polypeptide - TGAL (14696) F (complementing loci IHG, ITG; BR1A, BR1B)
In B/D segment (P)Blastogenic response to synthetic polypeptide - PHEGAL (14681) F (complementing loci BR3A, BR3B)
(P)Blastogenic response to synthetic polypeptide - GAT (14682) F (complementing loci IPHEG, IGAT;BR2A, BR2B)
(P)Blastogenic response to synthetic polypeptide - GLPHE (00000) F (comlementing loci BR4A, BR5B)
(P)Immune suppression (IS) (14685) H
Close to HLA-A Hemochromatosis (23520) LD, F
(P)Neutrophil differentiation factor (20270) LD
(L)F9 embryonic antigen (13701) H
Not close to HLA (P)Complement component C3b, receptor for: C3BR (12062) S
(P)Complement component C3d, receptor for: C3DR (12065) S
(P)Atrial septal defect, secundum type (10880) F
6p21.3-6p21.2(about 3cM from HLA) Glyoxalase I (13875) F, S
6p?linked to MHC (L)Corticosterone side-chain isomerase (12255) H
6p23 (P)Fragile site 6p23, folic acid type
6p (P)SALIVARY PROTEIN COMPLEX (16872 - 16881) F (See section IB.2 for details of this gene cluster.)

6p23-6q12 Prolactin (17674) REa (?between 6cen and GLO1)
6p23-6q12 (P)Insulin-like DNA sequence (14749) REa
6p23-6q12 (P)Initiator methionine tRNA (18062) REa (2 of 12+ genes on 6)
6q12 Malic enzyme, soluble (ME1) (15425) S
?between GLO1 and PGM3 Spinocerebellar ataxia-1 (16440) F - 15% male recomb./HLA
6q12 Phosphoglucomutase-3 (17210) S, F, OT
6q11-6q21 (P)Chorionic gonadotropin, alpha chain (shared with LH, FSH, TSH) (11885) REa
6q21-6q22 Oncogene: avian myeloblastosis virus, MYB (18999) S
6p23-6q12 (P)Oncogene: Kirsten rat sarcoma virus-1, KRAS1 (19011) S - pseudogene
6q21 Superoxide dismutase-2, mitochondrial (14746) S
6q26 Fragile site 6q26, type unknown
(L)Insulin dependent diabetes mellitus (22210) F, LD
(L)Manic-depressive illness (12548) F
(L)Pepsinogen (16970) F, H
(L)Paget disease of bone (16725) F
(L)Laryngeal adductor paralysis (15027) F
(P)Plasminogen activator (17337) S
Baboon M7 virus replication, BEVI (10918) S
(P)Monkey RBC receptor (15805) S
(I)Adenosine deaminase complexing protein-1 (10271) S
(P)Argininosuccinate synthetase pseudogene-2 (10784) REa
(others on 8 or more other chr. incl. X and perhaps Y)

In addition: 9 anonymous DNA segments, 1 antigen defined by a monoclonal, 1 surface antigen (18551), and 2 O'Farrell protein spots (HGM 7).

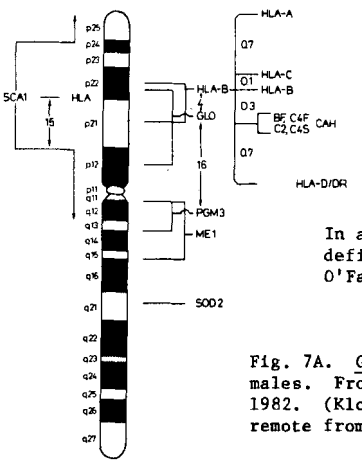


Fig. 7A. Gene map of chromosome 6. Recombination frequencies are for males. From Weitkamp and Lamm, HGM6, Cytogenet. Cell Genet. 32:133, 1982. (Klouda et al. (J. Med. Genet. 19: 337-341, 1980) placed CAH1 more remote from HLA-B, possibly outside the HLA complex, and close to DR.)

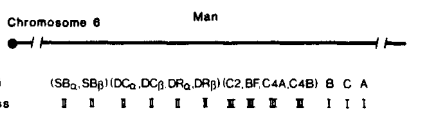


Fig. 7B. From Steinmetz and Hood: Science 222:727-733, 1983.

Chromosome No. 7

7p14-7pter (P)Gamma-glutamylcylotransferase (13717) S
7p21-7q22 (P)Argininosuccinate lyase (20790) S
7pter-7q22 Phosphoserine phosphatase (17248) S
7p14-7cen Biliverdin reductase (10975) S
Uridine phosphorylase (19174) S
?7p13(or 3p21.1) (L)Greig - polysyndactyly craniofacial dysmorphism syndrome (17570) Ch
7p22-7q22 Malate dehydrogenase, mitochondrial (15410) S
7p13-7p22(?p21-p22) Epidermal growth factor receptor (same as S6) (13155) S
7p11.2 Fragile site 7p11.2, folic acid type
7cen-7q22, Beta-glucuronidase (25322) S, D
7pter-7q22 (P)Oncogene ERBB (19014) REa
7q21-7q22 Collagen I (alpha-2) (12016) S, REa, D
(P)Collagen III (alpha-1) (12018) S
(P)Hydroxyacyl CoA dehydrogenase (14345) S
7q22-7qter Neutrophil migration (NM;granulocyte glycoprotein; GP130; formerly neutrophil chemotactic response, NCR) (16282) D
7q22-7qter (P)Trypsin-1 (27600) REA
7q22-7qter (P)Carboxypeptidase A (11485) REa —both serine proteases
7q21-7q36 HISTONE GENE CLUSTER (H1, H2A, H2B, H3, H4) (14271-14275) A
(P)Nonhistone chromosomal protein-1 (11887) S
(P)Growth rate controlling factor-1 (13922) S
(L)Diaphorase-2 (13560) S
(L)Colton blood group (11045) D (but see chr. 2)
7p11-7q11 (P)Asparagine synthetase (10837) S
?7p (L)Goldenhar syndrome (14140) Ch

In addition: 5 anonymous DNA segments, 1 surface antigen (18552), and 2 O'Farrell protein spots (H

Chromosome No. 8

(L)Fibronectin (?13560) S (?concerned with expression on cell surface)
8p11 (P)Spherocytosis (18290) F, Ch (linkage with t8;12 and t3;8; see chr. 14))
8p21.1 Glutathione reductase (13830) S, D
(P)Clotting factor VII expression (22751) D
8q23.3 Langer-Giedion syndrome (15023) Ch
?8q13-8qter (EM) (L)Glutamate-pyruvate transaminase, ?soluble liver (13822) S, EM (see chr. 16)
8q21.1-8qter (P)Glycine auxotroph B, complementation of hamster; GLYB (13848) S
8q22 Oncogene: Moloney murine sarcoma virus, MOS (19006) S
Fragile site 8q22.3, folic acid type
8q24.3 (L)Neoplastic lymphoproliferation-1 (16184) Ch
8q24 Oncogene MYC: avian myelocytomatosis virus (19008) A
(P)Tyroglobulin (18845) S
(P)Carbonic anhydrase II (11481) REa (2 forms of CA linked in Old World monkey and mouse)

ANLL, M2 = t(8;21) (q22.1; q22.3)

Burkitt and ALL, L3 = t (8;14) (q24.13; q32.33). Yunis, Science: 221:227, 1983. Burkitt also T(8;2) (q24;p12 t8;22)(q24;q11)

Chromosome No. 9

(P)Coproporphyrinogen oxidase (12130) S
9p13-9p24 Adenylate kinase-3, mitochondrial (10303) S, D
9p13-9p22 Aconitase, soluble (10088) S
9p13-9p21 Galactose-1-phosphate uridyltransferase (23040) S, D, F
(P)Fragile site 9p21.1, folic acid type
9p24-9p13 (distal to IFL)Interferon-F (beta or fibroblast interferon) (14764) REa, A
9pter-9p13 LEUKOCYTE INTERFERON GENE FAMILY (INTERFERON-L or alpha) (14766) REa, A
9qh (9q12) (P)Cytoplasmic membrane DNA (12633) A
9cen-9q34 (P)Folylpolyglutamate synthetase (13651) S
9cen-9qter (P)Glucose-regulated protein, GRP78 (13812) REa
9q32 Fragile site 9q32, folic acid type (13664)
9q34 Argininosuccinate synthetase (21570) S, D (pseudogenes on several chrs.)
(9q34) ABO blood group (11030) F
9q34 Adenylate kinase-1, soluble **(10300) F,S,D
Nail-patella syndrome (16120) F
Delta-aminolevulinate dehydratase (12527) F
Orosomucoid (13860) F

11
m*
v(NR)*
27
ORM-13-ALAD-11-AK-13-ABO
21
17

Linked to ABO definitely or provisionally. Linkage group assigned to No. 9 by assignment of AK1

(L)Dopamine-beta-hydroxylase (22336) F (lod 2.32, RF 0.0 with ABO)
(L)Waardenburg syndrome-1 (19350) F
(L)Multiple exostoses (13370)
***Onc gene: Abelson strain, murine leukemia virus (18998) REa, Ch
(P)Immunoglubulin epsilon heavy chain pseudogene: IGE3 (14721) A

In addition: 2 anonymous DNA segments and 4 O'Farrell protein spots (HGM 7)
*See section I,B for definitions.
Proximal to break in Ph^1 rearrangement (9q32.1).*Distal to 9q34.1.

Chromosome No. 10

10pter-10p11.1 Phosphofructokinase, platelet type (17184) S
10p11-10pter Hexokinase-1 (14260) S,D
?10q11-10q24 Adenosine kinase (10275) S, D-EM
10q11.1-10q24 Inorganic pyrophosphatase (PP) (17903) S, D
10q23 Fragile site 10q23 (folate-dependent) (13654)
10q25 Fragile site 10q25 (BrdU-dependent) (13662)
10q25.3-q26.1 Glutamate oxaloacetate transaminase, soluble (13818) S, D, H
(P)Glutamate-gamma-semialdehyde synthetase (13825) S
(P)Polykaryocytosis promoter (FUSE) (17475) S
(P)External membrane protein-130 (13371) S
?10q24-10q25 (P)Lysosomal acid lipase-A (27800) S, H
(I)Chorionic gonadotropin (11885) S (also see chr. 18; not structural locus)
(P)Oligomycin resistance (mitochondrial ATPase, ATPM) (16436) S
10q25.3-10q26.1 (P)Phosphoglycerate mutase A (17225) D,H

In addition: 2 anonymous DNA segments and 2 O'Farrell protein spots (HGM 7).

Chromosome No. 11

11pter-11q12	(P)Cathepsin D (11684) S
11p15	Insulin (17673) S, A, REb
11p13-11p15	(L)Beckwith-Wiedemann syndrome (13065) Ch (duplication)
11p13 (about 10 cM from NAG)	Catalase (11550) S, D
11p15.1-11p15.5	Oncogene HRAS1 (Harvey rat sarcoma-1) (19002) S (pseudogene HRAS2 on X)
11p1203-11p1208	Lactate dehydrogenase A (15000) S
11p11-11q12	Acid phosphatase-2 (17165) S
11p11-11q13	(P)Apolipoprotein A-I (10768) REa, RE
11p11-11q13	(P)Apolipoprotein C-III (23455) REa, RE (2.6kb 3' to APOA1), F
(11p11-11p13)	(L)Apolipoprotrein A-IV (10769) F
	(L)Apolipoprotein C-II (23455) F
11q13-11q22	Esterase-A4 (13322) S
	(P)Herpes virus sensitivity (14246) S (see chr. 3)
11p13	Wilms tumor/aniridia/gonadoblastoma/retardation (WAGR) complex (19407) (
11p13	(L)Aniridia-2 (10620) Ch
	(P)BVIX (BALB virus induction, xenotropic) (11399) S
?11p	(L)Tyrosinase-negative albinism (20310) H — Based on
	(L)Malic enzyme, mitochondrial (15427) H — mouse map
11p15	NON-ALPHA GLOBIN CLUSTER (NAG; Hemoglobin beta cluster; HBBC)
(polarity: cen-HBE-HBG-HBD-HBB-pter)	Hemoglobin epsilon (14210)-AAS
	Hemoglobin gamma 136 glycine (14225)
	Hemoglobin gamma 136 alanine (14220)
	(P)Hb gamma regulator (14227) RE — S, HS, REa
	Hemoglobin delta (14200) — AAS
	Hemoglobin beta (14190) LD,AAS,F
	Hpa I recognition polymorphism (14302)RE
about 15cM from HBB	F-cell production (heterocellular hereditary persistence of fetal hemoglobin) (14247) F
Proximal to HBBC	Parathyroid hormone (16845) REa
11q13.3	Fragile site 11q13 (13655)
11p13-11q22	Glutathione S-transferase-3 (formerly called GST1) (13835) S
11q13-11pter	(P)Neutral alpha-glucosidase AB (10416) S
11q23.2-11qter	Porphobilinogen (PBG) deaminase (uroporphyrinogen I synthase) (17600)S
11q23.3	(P)Fragile site 11q23.2 (folic acid type)
	(L)Fibronectin (?13560) S (?fibrillar morphology of cell FN)
	(P)Leu-7 (HNK-1) antigen of natural killer lymphocytes (15129) S
	(L)Congenital glaucoma-1 (23130) Ch
	(P)Collagenase (recessive epidermolysis bullosa dystrophica) (22660) S
	(P) 2',5'-oligoisoadenylate synthetase (16435) S

Order: Cen--PTH-11-15cM-HBBC-8cM-HRAS1-4-7cM-INS-pter

In addition: 13 anonymous DNA segments, 5 surface antigens (15125, 15126, 15127, 18554, 29 antigens, not necessarily all separate and different, defined by monoclonal antibodies (e.g., 14304, 15806, 15807), and 1 O'Farrell protein spot (HGM 7).

*See Mendelian Inheritance in Man, 6th ed., 1982) for amino acid substitutions in gamma, delta and beta variants and for molecular changes in deletion and nondeletion forms of beta-thalassemia.

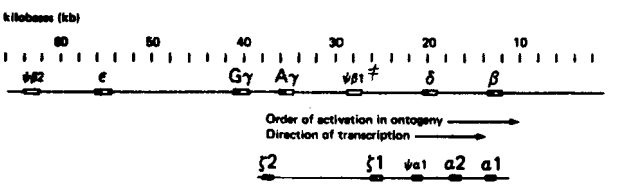


Fig. 8A. The linkage arrangement of human beta-like and alpha-like globin genes. The top line shows the relative locations of the five functional beta-like globin genes and the two beta-like pseudogenes. The bottom line shows the map of the four functional alpha-like globin genes and the alpha-globin pseudogene "psi alpha 1". The mRNA coding sequences are designated by filled rectangles. From Proudfoot, et al.: Science 209:1329, 1980. Shen and Smithies (Nucl. Acids Res. 20:7809, 1982) could not identify globin-like sequences in region labeled psi-beta-2.

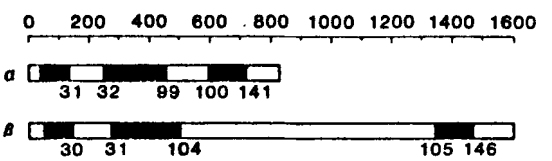


Figure 8B. The coding (shaded) and noncoding segments of the alpha and beta globin genes. (From Proudfoot et al., Science 209:1329, 1980.)

Chromosome No. 12

(P)Methionyl-tRNA synthetase (16656) S
(P)Alpha-glycerophosphate dehydrogenase (13846) S
(P)Phenylalanine hydroxylase (26160) REa
12pter-12q12 Branched chain amino acid transferase-1(11352) S
12p (P)Aromatic alpha-keto acid reductase (10792) S
12pter-12q14 (P)Oncogene INT1 (murine mammary cancer virus) (16482) REa
12p13 *Glyceraldehyde-3-phosphate dehydrogenase (13840) S, D, R
12p13 Triosephosphate isomerase-1 (19045) S, D, R
(L)Triosephosphate isomerase-2 (19046) S
(P)Collagen, alpha-1(I)-like (COLM) (12011) REa
?12p12.1,??12q24.2 Oncogene ras2 (KRAS2): Kirsten rat sarcoma virus (19007) S
12p121-12p122 Lactate dehydrogenase B (15010) S, D
(L)Lactate dehydrogenase C (15015) H
12p11-12p12 Enolase-2 (13136) S
12p11-12qter Citrate synthase, mitochondrial (11895) S
12q12-12q14 Serine hydroxymethyltransferase (13845) S, R
12q13.1 Fragile site 12q13.1, folic acid type (13663)
(L)Human leukocyte antigen: MIC3 (?distinct from S8) (14303) S
12q21 Peptidase B (16990) S
12q21-12q24.2 (L)Familial intestinal neurofibromatosis (16222) Ch
12q24.1 Interferon, gamma or immune type (14757) S, A (3 introns; IFF, IFL none)
(P)Elastase-1 (13012) REa
(P)UV-damage, excision repair of (XP complementation group I) (19206) S

Also: 3 DNA segments, 1 surface antigen (18556), 3 antigens by monoclonals, 3 spots (HGM7).
*Probable order: 12pter-TPI-GAPD-LDHB-ENO2-cen-SHMT-PEPB-12qter.

Chromosome No. 13

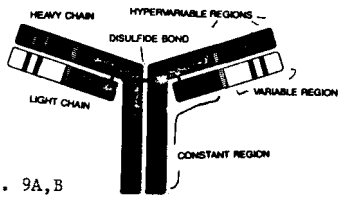
13p12 Ribosomal RNA (18045) A
13q14 Esterase D (13328) S, F, D
13q14 Retinoblastoma-1 (18020) Ch
13q14 (L)X-ray sensitivity (19443) Ch
(L)Dubin-Johnson syndrome (23750) LD (with factor VII deficiency)
13q34 (P)Clotting factor VII (22750) D
13q34 (P)Clotting factor X (22760) D
?13q34 (L)Carotid body tumor (16800) F (?linked to factors VII and X)
13q14-13q31 (L)Letterer-Siwe disease (24640) Ch
(P)UV-damage, excision repair of (Xp complementation group I) (19206) S

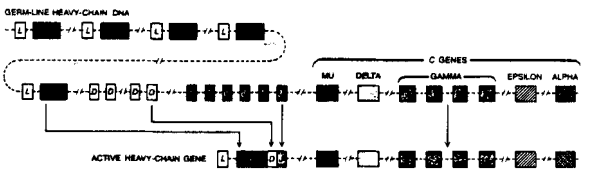
In addition: 7 anonymous DNA segments (HGM 7).

Chromosome No. 14

14p12 Ribosomal RNA (18045) A
Lentil agglutinin binding (LCH) (15103)S
14q12.00-14q13.105 Nucleoside phosphorylase (16405) S, D
14q21-14qter Tryptophanyl-tRNA synthetase (19105) S
(P)External membrane protein-195 (13374) S
14q32 Creatine kinase, brain type (12328) S
14q22-14qter (P)Phosphoribosylglycineamide formyltransferase (17246) S
(P)Phosphoribosyl formylglycinamidine synthetase (10258) S
(P)Esterase activator (13325) S
?1 multifunctional protein involved in de novo purine synthesis
?linked to IGHC (L)Spherocytosis, Denver type (18290) F
(L)T-cell idiotypic receptor for MHC antigens (18688) H
14q24.3-q32.2 Protease inhibitor (PI; alpha-1-antitrypsin) (10740) F, S
14q24.3-14q32.1 DNA segment D14S1 (10775) REa, A (2 other anonymous DNA segments - HGM7)
About 20cM apart IMMUNOGLOBULIN HEAVY CHAIN GENE FAMILY REa, A **
Variable region genes (about 250) (14707)
D (for diversity) region genes (many) (14708)
J (for joining) region genes (more than 4) (14701)
Constant region of heavy chain of IgM1 (14702)
Constant region of heavy chain of IGM2 (14703)
Constant region of heavy chain of IgD (14717)
Constant region of heavy chain of IgG2 (14713)
(14q32.33-14qter) Constant region of heavy chain of IgG4 (14711)
5'- G2-17kb-G4 - 3'
Constant region of heavy chain of IgG3 (14712)
Prob. orientation: Constant region of heavy chain of IgG1 (14710)
close linkage known from Lepore-like myeloma protein
cen-PI-D14S1-IGHC- Constant region of heavy chain of IgE (14718)
-IGHV-ter (3' centromeric; Constant region of heavy chain of IgEP1 (14719)
IgEP2 on chr. 9

5', telomeric; IgM Constant region of heavy chain of IgA1 (14690)
telomeric to IgG). Constant region of heavy chain of IgA2 (14700)
**A Tunisian deletion indicates order: 5' - G3 - G1 - psi E1 - A1 - G2 - G4 - E - A2 - 3' (Lefranc et al., Nature 300: 760, 1982). 5' - E2 - E1 - E3 - 3' (Nishida et al., PNAS 79:3833, 1982; E3 = ?pseudogene). Following information from J. J. Johnson and L. L. Cavalli-Sforza (Stanford Univ., Nov., 1983: 5'(qter)---V--(7cM)--D--J--8kb--mu--5kb--delta--?--gamma-3--26kb--gamma-1--19kb--pseudo-epsilon-1(pseudo-epsilon-2 on chr. 9)--13kb--alpha-1--?--gamma-2--18kb--gamma-4--23kb--epsilon--10kb--alpha-2----3'(centromere).





From Leder, P.: Scientific American 246 (May):102, 1982.

Fig. 9A,B

Chromosome No. 15

15p12	Ribosomal RNA (18045) A
15q11-15q13	Alpha-mannosidase-A, cytoplasmic (15458) S, D
15q11	Prader-Willi syndrome (17627) Ch
?near centromere	(P)Dyslexia-1 (12770) Fc
15q11-15qter	(P)Coronavirus 229E sensitivity (12246) S
15q21-15q22	Beta-2-microglobulin (10970) S, D, H
15q22-15qter	Mannosephosphate isomerase (15455) S
15q22-15qter	Pyruvate kinase-3 (PKM2) (17905) S,D
15q22-15q25.1	Hexosaminidase-A (27280) S
15q21-15qter	Isocitrate dehydrogenase, mitochondrial (14765) S
	(P)BVIN (BALB virus induction, N-tropic) (11398) S
	(P)Neutral alpha-glucosidase C (10418) S
15pter-15q21	Sorbitol dehydrogenase (18250) S, H
15q25-15q26(?15q26)	Oncogene: feline sarcoma virus (19003) S, A
15q11-15qter	(P)Actin, cardiac alpha (ACTC) (10254) REa

In addition: 1 anonymous DNA segment, 2 antigens defined by monoclonal antibodies, and 2 O'Farrell protein spots (HGM 7).

Chromosome No. 16

5cm from GPT1	(P)Macular dystrophy, atypical vitelliform (15384) F (on 16 if GPT1 is)
?16p11-16pter	(I)Glutamate pyruvate transaminase, ?soluble red cell (13820) S (see chr. 8)
	(P)Epidermolysis bullosa, Ogna type (EBS1) (13195) F (c,F)*
	(P)Cystathionase (21950) S
16p12-16pter	(L)Hb H mental retardation syndrome (14175) F
	ALPHA GLOBIN GENE CLUSTER****
16p12-16pter	Hemoglobin zeta (2 loci) (14230,14231) RE — RE,A,D**
16p12-16pter	Hemoglobin alpha (1, 2, or 3 loci) (14180) S, HS — RE,A,D**
16p12-16p13	Phosphoglycolate phosphatase (17228) S
16q12-16q21	Diaphorase-4 (12586) S
16p12.3	Fragile site 16p12.3, folic acid type (13656)
16p12-16q22	Glutamate oxalacetic transaminase, mitochondrial (13815) S
Order: pter-PGP-0.25-16qh-0.17-GOT2-0.08-HP-qter (Jeremiah et al., loc.cit.,1982)	
16q12-16q22***	Adenine phosphoribosyltransferase (10260) S, D
?linked to HP	(L)Congenital cataract (11559)
16q22	Haptoglobin (14010) Fc — v*
16q22	Lecithin-cholesterol acyltransferase (24590) F, LD — v*
16q22.1	Fragile site 16q22.1, distamycin A type (13657)
16q23	Fragile site, 16q23 (type unknown)
	(P)Chymotrypsinogen B (11889) REa - 19% AA homology to HP
	(P)Thymidine kinase, mitochondrial (18829) S
	(P)Lysosomal acid lipase-B (24798) S
	(P)Growth rate controlling factor-2 (13923) S
	(P)Esterase-B3 (13329) S
	(P)Glyoxalase II (hydroxyacyl glutathione hydrolase; HAGH) (13876) S
	(P)Nonhistone chromosomal protein-2 (NHCP-2) (11888) S

(P)Antiviral state repressor regulator (interferon regulator) (10747) D
(P)Vesicular stomatitis virus defective interfering particle repressor (12602) S
(L)Transcobalamin II (27535) H (or perhaps chr. 17)

In addition: 2 anonymous DNA segments (HGM 7).
*See section I,B for definition.
**See Figs. 6 A, B, and C.
***Distal to GOT2 and DIA4 (Jeremiah et al., Ann. Hum. Genet. 46:145, 1982)

Chromosome No. 17

?17p13 (L)Miller-Dieker lissencephaly syndrome (24720) Ch
17pter-17p11 (P)MYOSIN, SKELETAL, HEAVY CHAIN (16073) REa (myosin gene cluster also on 7)
MYHSA1 = myosin heavy chain, adult-1 (16073) REa
MYHSA2 = myosin heavy chain, adult-2 (16074) REa
MYHSE1 = myosin heavy chain, embryonic-1 (16072) REa
17p12 Fragile site 17p12, distamycin A type (13665) Ch
17q210-17q220 Galactokinase (23020) S,Ch,R,C
17q210-17q220 GROWTH HORMONE/PLACENTAL LACTOGEN GENE CLUSTER S, REa, A, C
(17q22-17q24 by in situ hybridization. By CMGT, order = cen-GALK-GH-[TK-COL1A1]
GHN = Growth hormone, normal (13925) S, REa, A
CSL = Chorionic somatomammotropin-like (15020) S, REa, A
CSA = Chorionic somatomammotropin A (15020) S, REa, A
GHV = Growth hormone variant (13925) S, REa, A
CSB = Chorionic somatomammotropin B (15020) S, REa, A
5' → 3' about 45 kb
17q21-17q22 (A)(closer to TK than GALK) Collagen I alpha-1 polypeptide (12015) S, M, A, REa
(L)Collagen IV alpha-1 polypeptide (12013) REa
17q210-17q220 Adenovirus-12 chromosome modification site-17 (10297)V
17q210-17q220 Thymidine kinase-1 (18830) S, Ch, R, C
17q22-17q25 Acid alpha-glucosidase (23230)S
(L)Creatine kinase, brain type (12328) S
17p11-17q21 Oncogene: avian erythroblastic leukemia virus, ERBA (19012) REa

In addition: 1 anonymous DNA segment, 1 antigen defined by a monoclonal antibody (15656), and 1 surface antigen (18557) (HGM 7).

Chromosome No. 18

18q23 Peptidase A (16980) S, D
18p11 (I)Human chorionic gonadotropin, alpha subunit (11885) S, A (see chr. 10)
18q24.3 (L)Neoplastic lymphoproliferation-2 (16185) Ch [t(14;18) in non-Hodgkin lymphoma]
(P)Oncogene: endogenous retrovirus-1, ERV1 (13115) REa
(P)Asparaginyl-tRNA synthetase (10841) REa

In addition: 3 anonymous DNA segments (HGM 7).

Chromosome No. 19

CHORIONIC GONADOTROPIN, BETA CHAIN (at least 8 genes) (11886) REa
Luteinizing hormone, beta chain (15278) RE
Beta chains of FSH, TSH probably linked. CGB, LHB, FSHB, TSHB = Glycopeptide hormone gene cluster.
19pter-19q13 Lysosomal alpha-D-mannosidase-B (24850) S
19p13.2-19q13 Glucosephosphate isomerase (17240) S, D
19q Polio virus sensitivity (17385) S
19q (L)Echo 11 sensitivity (12915) S
19pter-19q13 Peptidase D (17010) S, F
(P)Beta-glucuronidase, mouse, modifier of (23161) S
(P)Baboon M7 virus receptor (?same as virus RD114 receptor) (10918) S
(P)Coxsackie B3 virus susceptibility (12005) S
(P)Branched chain amino acid transaminase-2 (11353) S
19pter-19q13 (P)Lysosomal DNA-ase (12635) S
Linked to C3(@25cM) Familial hypercholesterolemia (LDL receptor) (14389) F, S
Probable order: Complement component-3 (12070) S (Le@ 7 cM in males vs. C3 RFLP)
FHC-C3-Le-DM-Se-Lu Lewis blood group (11110) F (linked to serum C3)
Linked to PEPD Myotonic dystrophy (16090) F (in C3-Le-Se-Lu linkage group)
Secretor (18210) F
@13cM from C3 (P)Apolipoprotein E (20776) F (?close to Se)
Lutheran blood group (11120)
(P)Bombay phenotype (Hh) (21110) —(v,F)* LE-C3-DM-(Se-PEPD)-Lu
Order suggested by Eiberg et al. Clin. Genet. 24: 159, 1983
Linked to DM (P)Neurofibromatosis (16220) F
(P)Ferritin, heavy chain (13477) S
(P)Ferritin, light chain (13479) S

In addition: 3 anonymous DNA segments (HGM 7).
*See Section I,B for definition.

Chromosome No. 20

20p	Inosine triphosphatase (14752) S
20p11.23	Fragile site 20p11.23, folic acid type (13658)
20p12.2	(P)Multiple endocrine neoplasia, type 2 (17140) Ch
20q132-20qter	Adenosine deaminase (10270) S, D, REa
20cen-20q13	S-adenosylhomocysteine hydrolase (18089) S
	(P)Desmosterol-to-cholesterol enzyme (DCE) (12565) F
	Protooncogene src (Rous sarcoma) (19009) REa

In addition: 4 anonymous DNA segments (HGM 7).

Chromosome No. 21

21p12	Ribosomal RNA (18045) A
21q21-21qter	Antiviral protein (interferon receptor) (10745) S, D
21q22.1	Superoxide dismutase-1 (soluble) (14745) S, D
21q22	Phosphoribosylglycinamide synthetase (13844)S, H ┐ ?1 multifunctional prot
	(P)Phosphoribosylaminoimidazole synthetase (17244) S ┘ involved in de novo
	(P)Surface antigens 14 (18559) S purine synthesis
21q22	Phosphofructokinase, liver type (17186) S, D
21q11-qter	(P)Primary thrombocytosis (18795) Ch
	(L)Beta-amino acids, renal transport of (10966) D
	(L)Beta-adrenergic stimulation, response to (10967) D
	(P)5-hydroxytryptamine oxygenase regulator (14346) D
	(P)Cystathionine beta-synthase (23620) S

In addition, 9 anonymous DNA segments, 1 surface antigen (18559) and 2 antigens defined by monoclona (HGM 7).

Chromosome No. 22

22p12	Ribosomal RNA (18045) A
22pter-22q11	(L)Cat eye syndrome (11547) Ch
22q11	DiGeorge syndrome (18840) Ch
?22q11.12	*IMMUNOGLOBULIN LAMBDA LIGHT CHAIN GENE FAMILY* REa, A
6 or more constant region genes in tandem, each with J gene	Variable region of lambda light chains (many genes) (14724)
	J region of lambda light chains (several genes) (14723)
	Constant region of lambda light chains (14722)
22q11.3 (distal to lambda)	Chronic myeloid leukemia (15141) Ch [Ph^1=t(9;22)(q34.1;q11.21)]
22q13.31-22qter	**Arylsulfatase A (25010) S
22q13.31-22qter	NADH-diaphorase-1 (25080) S
22q11-22q13	**Aconitase, mitochondrial (10085) S
22q13	**N-acetyl-alpha-D-galactosaminidase (alpha-galactosidase B) (10417) S
22q13-22qter	Beta-galactosidase-2 (10968) S
22pter-22q11(Ph^1)	(P)Alpha-L-iduronidase (25280)S, D
22q11-22qter	Oncogene SIS: simian sarcoma virus (19004) S

In addition: 7 anonymous DNA segments and 1 surface antigen (18558) (HGM 7).
**Distal to the break point that creates Philadelphia chromosome.

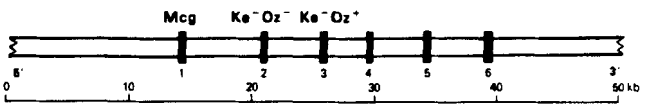


Fig. 10. The lambda constant region genes are 6 in number located in a 30 kb segment; 3 of them correspond to 3 of the 4 known nonallelic lambda constant chains. At least 3 other lambda constant genes have not been positioned. One of these presumably has the Kern(+)Oz(-) characteristic. The amino acid differences of these nonallelic forms is known. (Hieter et al., Nature 294:536-540, 1981.)

B. Linked autosomal loci for which assignment to a specific chromosome has not yet been achieved.

The tightness of the linkage is stated in general terms defined as follows:

v - very close - recombination less than 2% (NR - no recombinant observed)
c - close - recombination 2 - 6%
m - medium - recombination 6 - 22%
l - loose - recombination more than 22%
? - lods 2.0-3.0

1. Phenylthiocarbamide taste (PTC) locus (17120)—(c, F) (See inconsistent data of Neiswander et al., Cytogenet. Cell Genet. 32:303, 1982.)
   Kell blood group (K) locus (11090)—

2. SALIVARY PROTEIN COMPLEX (SPC)
   Proline rich parotid salivary protein (Pr) (16878)—
   Parotid acidic protein (Pa) (16873)—(c,F,LD)
   Double band parotid salivary protein (Db) (16877)—(?,m,F,LD)
   Parotid salivary glycoprotein (Gl) (16880) - Linkage disequilibrium
   Probable order: Pa - Pr - Db - Gl
   PIF (parotid isoelectric focusing) protein (16872)
   (Ps and PIF closer to Gl and Db than to Pr or Pa)
   Parotid middle band protein (Pm) (16878)— linked to above five loci
   Parotid size variant (Ps) (16881)—
   Parotid basic protein (Pb) (16875)— linkage unknown
   Post-parotid basic protein (PPb) (16876)—
   CON1 (16887) - closely linked to Ps
   CON2 (16888) - closely linked to PmF
   (SPC is probably linked to GLO1 and Bf and therefore on 6p; confirmation by linkage to HLA and by use of recombinant DNA probes is awaited.)

3. Five lipoprotein-Ag loci (15200) (?v,F)

4. Complement component-6 (C6) (21705)—(? v,F) Linked in dog, ?marmoset. In man, genes physically close by DNA studies (RE).
   Complement component-7 (C7) (21707)— Linkage supported by observation of combined deficiency (Lackmann, Clin. Exp. Immun. 33:193, 1978).

5. Ii blood group (11080)—
   Congenital cataract (21250)—(?c,F)

6. Epidermolysis bullosa progressiva, EBR3 (22650)—
   Hypoacusis (HOAC; a recessive partial deafness) (22070)—(? c, F)
   ?Red hair (26630)

7. Cerebellar ataxia, a recessive form (21320)—
   Tyrosinase-negative albinism (20310)—(?c,F) From homology to tyrosinase-negative albinism in the mouse, which is linked to NAG, these genes may be on 11p in man.

8. Marinesco-Sjogren syndrome (25880)—
   Hypergonadotropic hypogonadism (21285)—(m,F) Lod score more than +30.

In addition, the chromosomal location of the GPT1 linkage group is not completely certain with assignment claimed particularly for chromosomes 8 and 16 but also for chromosome 10 and very weakly for chromosome 13 (HGM7). This linkage group is as follows:
Glutamate pyruvate transaminase, soluble red cell GPT1 (13820)
Macular dystrophy, atypical vitelliform, VMD1 (15384) - 5cM from GPT1
Epidermolysis bullosa, Ogna type, EBS1 (13195) - closely linked to GPT1
(Linkage of breast cancer, 21200, to GPT1 unsubstantiated and perhaps disproved.)

For a collection and collation of published linkage data, see the following:
Keats, B.J.B., Morton, N.E., Rao, C.C, and Williams, W.R.: A Source Book for Linkage in Man. Baltimore: Johns Hopkins University Press, 1979; Keats, B.J.B.: Linkage and Chromosome Mapping in Man. Honolulu: University Press of Hawaii, 1981.

C. Genes known to be linked from DNA mapping but chromosomal location unknown.

1. tRNA(gln) - 18063
   tRNA(lys) - 18064

II. GENE MAP OF THE Y CHROMOSOME

(See Buhler: A synopsis of the Y chromosome. Hum. Genet. 55:145-175, 1980.)

A. From the study of variant Y chromosomes, a factor (or factors) which determines the differentiat of the indifferent gonads into testes is known to be located on the Y chromosome, probably on the short arm; this may be called testis determining factor (TDF). (See Davis, J. Med. Genet. 18:161-195, 1981.)

B. Histocompatibility antigens determined by the Y chromosome were first found in the mouse (Eichwa E.J., and Silmser, C.R.: Transplant Bull. 2: 148-149, 1955; see review by Gasser, D.L. and Silver, W.K.: Genetics and immunology of sex-linked antigens. Adv. Immun. 15: 215-217, 1972) and are now kn also in the rat, guinea pig, and many other species. Their existence in man was first shown by the fact that mouse antisera react with human male
lymphocytes but not with female lymphocytes (Wachtel, S.S., Koo, G.C., Zuckerman, E.E., Hammerling, U., Scheid, M.P., and Boyse, E.A.: Serological cross reactivity between H-Y (male) antigens of mouse and man. Proc. Nat. Acad. Sci. 71: 1215-1218, 1974). The strong likelihood that the locus that determines heterogametic sex determination and that for the H-Y antigen are one and the same was suggested by Wachtel et al. (Possible role for H-Y antigen in human males with two Y chromosomes. New Eng. J. Med. 293: 1070-1072, 1975.) The hormone-like action of the H-Y antigen in the bovine freemartin was discussed by Ohno et al. (Nature 261: 597, 1976). See also Wachtel et al., New Eng. J. Med. 295: 750, 1976. H-Y and TDF may be one and the same. Like HLA, H-Y is a differentiation antigen. (See Wachtel: Where is the H-Y structural gene? Cell 22: 3-4, 1980.) Work by Ohno using recombinant DNA probes for H-Y suggests that the Y-linked locus is regulatory, not structural, and that the structural locus, an ancient homology, is on Xp.

C. The existence of factors controlling spermatogenesis on the nonfluorescent part of the long arm of Y (distal part of Yq11) was suggested by study of 6 men with deletion of this segment and azoospermia (Tiepolo, L. and Zuffardi, O., Hum. Genet. 34: 110-124, 1976). This has been called azoospermia third factor (symbolized Sp-3).

D. That one or more genes concerned with stature are on the Y chromosome is suggested by the comparative heights of the XX, XY and XYY genotypes; that the effect of the Y chromosome on stature is mediated through a mechanism other than androgen is suggested by the tall stature of persons with XY gonadal dysgenesis (30610). See also the argument, from XO and XXY cases, that genes determining slower maturation must be on the Y (Tanner, J.M., Prader, A., Habich, H. and Ferguson-Smith, M.A.: Genes on the Y chromosome influencing rate of maturation in man. Skeletal age studies in children with Klinefelter's (XXY) and Turner's (XO) syndromes. Lancet II: 141-144, 1959). The postulated locus is symbolized STA (for "stature"). Yamada et al. (Hum. Genet. 58:268-270, 1981) found a correlation between the length of heterochromatic band Yq12 and height.

E. Alvesalo and de la Chapelle (Ann. Hum. Genet., 43:97-102, 1979; GMC-5, Edinburgh, 1979) suggested, on the basis of tooth size in males of various Y chromosome constitutions, that a Y-chromosomal gene controlling tooth size is independent of the testis-determining gene and is carried by Yq11 (symbolized TS for "tooth size"). See Alvesalo and Portin, Am. J. Hum. Genet. 32: 955-959, 1980.

F. HGM7 (Los Angeles, 1983) catalogued 10 single copy DNA segments mapped to the Y chromosome.

G. Repetitive sequences located exclusively or predominantly to the Y chromosome (e.g., Kunkel, Smith, Boyer, Biochemistry 18: 3343-3353, 1979) map to the heterochromatic portion of Yq and are presumably genetically inert because persons lacking these are phenotypically normal and normally fertile.

H. An argininosuccinate synthetase pseudogene is on the Y (Daiger et al., Nature 298:682, 1982).

I. The only structural gene confidently identified on the Y chromosome is that homologous to the X-linked gene for surface antigen MIC (Goodfellow et al., Nature 302:346, 1983)i.e., this is the sole known Y-specific gene product. (See Fig. 1 of Burgoyne, Hum. Genet. 61:95, 1982, for suggested homologous segment of X and Y.)

III. GENE MAP OF THE X CHROMOSOME

From the characteristics of X-linked inheritance and in two instances from studies of somatic cell hybrids, over 115 genetic loci have been assigned to the X chromosome. Those assigned by somatic cell genetics include tyrosine aminotransferase regulator on Xq (TATr, 31435), surface antigen, X-linked (SAX, 31345), temperature sensitive complement, C1HR (31365), monoamine oxidase A (30985), an actin gene (30004), a beta-tubulin gene (31434), ouabain sensitivity (31135) and a pseudogene for argininosuccinate synthetase (21570). (The first of these has been disputed.) The others were assigned by pedigree pattern (in the first instance, at least); confirmation by other methods, such as study of cell hybrids and mapping to the X chromosome in other mammals (Ohno's law), is available for some. In yet other instances (Lesch-Nyhan syndrome, testicular feminization, ornithine transcarbamylase deficiency, PRPP synthetase, etc.) confirmation was provided by demonstration of lyonization in cell cultures or other material from heterozygotes. Actin genes (30005) have been identified on both the X and the Y (J.L. Mandel, Strasburg). The HRAS-2 oncogene, a pseudogene, is on the X (S. J. O'Brien et al., Nature 302: 839, 1983).

A. Regional assignments on the X chromosome by study of cell hybrids.
(Bracket on left indicate assignment by methods other than cell hybridization)

[Xp22.3	Xg blood group (31470)
[Xp22.3	Steroid sulfatase (ichthyosis) (30810)
[(L)Xp22	Aicardi syndrome (30405) Ch (balanced translocation)
[(L)Xp22.32	Chondrodysplasia punctata (30295) Ch
[(L)Xp21-Xp22	Gonadal dysgenesis, XY female type (30610) (?deficiency of gonad-specific H-Y receptor)
[(P)Xp223	H-Y regulator (or repressor) (30697) — [May be the same. (See II,B for reference to structural H-Y locus on Xp.)
Xp21-Xp223	Polymorphic lambda RC8 probe about 10 cM from DMD and 15cM from RS (DXS9)
[Xp21	Duchenne muscular dystrophy (31020) Ch, F
[?Xp21	Becker muscular dystrophy (31010) Fd ?Ch ?allelic
Xp21 (prox. to DMD)	Ornithine transcarbamoylase (31125) Ch
Xp11-Xq11 (Xq25)	Actin (30002) REa - ?pseudogene
Xp110-Xp113	Polymorphic DNA probe L1.28 about 15 cM from BMD (DXS name)
(P)Xp11-Xq13	Testicular feminization (31370) S — [Closely linked
Xp11-Xq13	Menkes syndrome (30940) H — [in mouse
Xp11-Zq11 (X125)	Actin (30002) REa - ?pseudogene
[(L)Xq112-Xq211	X chromosome controlling element (31467)
[(L)Xq12	Aarskog-Scott syndrome (30540) Ch
[(L)Xq12	Anhidrotic ectodermal dysplasia (30510) F
Xq13	Phosphoglycerate kinase (PGK) (31180)
Near PGK	Monoamine oxidase A (30985)
Xq	Mucopolysaccharidosis II (30990) Ch
Xq22-Xq24	Alpha-galactosidase A (Fabry disease) (30150)
Xq22-Xq26	Phosphoribosylpyrophosphate synthetase (31185)
Xq26-Xq27	Hypoxanthine-guanine phosphoribosyltransferase (30800) - about 35 cM from FS
near HPRT(Xq13-Xq27)	Temperature sensitivity, mouse and hamster, complement; BA2R (31365)
Xq27-Xq28	Hemophilia B (factor IX) (30690) REa, A, F, D - no recombination with FS
Xq27.3	Fragile site Xq27.3 (at interface between Xq27 and Xq28)(30955)*
Xq28	Clucose-6-phosphate dehydrogenase; G6PD (30590) S
Xq28(distal to G6PD)	Goeminne TKCR syndrome (31430) Ch (balanced translocation)

*A second X chromosome fragile site is located at Xq26.

Radiation-induced segregation (Goss-Harris) supports order:

PGK - GALA - PRPS - HPRT - SAX - G6PD.

B. Regional mapping by family linkage studies

1. The Xg cluster (linkages to Xg)

Xg (31470)**
Xp22 Ichthyosis (steroid sulfatase deficiency) (30810) - m
v — [Chronic granulomatous disease (30640) - c* — [Xk (31485) - v — ?same locus
Ocular albinism, Nettleship-Falls type (30050) - m

(P)Ocular albinism, Forsius-Eriksson type (30060) - m
Retinoschisis (31270) - l (about 25cM from Xg)
(L)Mental retardation, ? type (30953) - m
**Ropers et al (HGM6) suggested order: Xg - STS - Xk - cen.
Ferguson-smith (HGM6) suggested order: STS - 11cM - Xg - ?2cM - Xk - OA.

2. The G6PD cluster (linkages to G6PD unless otherwise indicated)

Xq28 Glucose-6-phosphate dehydrogenase (30590) - about 6cM from FS
Deutan colorblindness (30380) - c
Protan colorblindness (30390) - c
Hemophilia A ((30670) - c
Adrenoleukodystrophy (30010) - v
(L)Blue-monochromatic colorblindness (30370) - c
(L)Manic-depressive psychosis (30920) - c
Xm (31490) - m (linked to colorblindness)
(L)Emery muscular dystrophy (31030) - m (? linked to colorblindness)
(P)Hunter syndrome (30990) - m (? linked to Xm) - ?inconsistent with information above.
Xq27-q28 Mental retardation with large testes and fragile site (30955)
(Linkage to G6PD or colorblindness has been demonstrated;in turn linked closely to HEMB but HEMB not linked to the G6PD cluster.)

* See Section I, B for definitions of v, c, m, l.
(See assemblage of X-linkage data in Race and Sanger, Blood Groups in Man, 6th edition, 1975.)

C. In addition, 77 single copy anonymous DNA segments useful in mapping and some polymorphic have been assigned to the X chromosome and almost all have been mapped to a specific region of X (HGM 7, Los Angeles, 1983). The X and Y chromosomes share 11 single copy DNA probes, all located on Xq12-Xq (HGM 7). By O'Farrell 2-D electrophoresis, 7 function-unknown protein spots coded by the X chromoso have been identified (HGM 7). Also, the genes for 4 cell antigens identified by monoclonal antibodi and 2 surface antigens (e.g., 31345) have been mapped to the X chromosome (HGM 7). At least 2 X-chromosome-specific repetitive DNAs of unknown function have been found.

Table II

Number of loci identified mainly by mendelizing phenotypes*
From McKusick: Mendelian Inheritance in Man, 6th ed., 1983.

Phenotype	Verschuer 1958**	Mendelian Inheritance in Man						
		1966 (1st ed.)	1968 (2nd ed.)	1971 (3rd ed.)	1975 (4th ed.)	1978 (5th ed.)	1982 (6th ed.)	Sept. 29 1983
Autosomal Dominant	285	269(+568)	344(+449)	415(+528)	583(+635)	736(+753)	934(+893)	996(+959)
Autosomal Recessive	89	237(+294)	280(+349)	365(+418)	466(+481)	521(+596)	588(+710)	599(+741)
X-Linked	38	68(+51)	68(+55)	86(+64)	93(+78)	107(+98)	115(+128)	118(+137)
Total	412	574(+913)	692(+853)	866(+1010)	1142(+1194)	1364(+1447)	1637(+1731)	1713(+1837)
Grand Total	412	1487	1545	1876	2336	2811	3368	3550

*Numbers in parenthesis refer to loci not yet fully identified or confirmed.
**Lehrbuch der Humangenetik (Munich: Urban and Schwartzenberg, 1958).

For methodologic and interpretive discussions of the human gene map, see the following references:

1. McKusick, V.A. and Ruddle, F. H.: The status of the gene map of the human chromosomes. Science 196:390-405, 1977.

2. McKusick, V.A.: The anatomy of the human genome. (Wilhemine E. Key Lecture) J. Hered. 71:370-391, 1980.

3. Ruddle, F. H.: A new era in mammalian gene mapping: somatic cell genetics and recombinant DNA methodologies. Nature 294:115-120, 1981.

4. McKusick, V. A.: The human genome through the eyes of a clinical geneticist. Cytogenet. Cell Genet. 34:7-23, 1982.

Other useful references:

Land, H., Parada, L.F., and Weinberg, R. A.: Cellular oncogenes and multistep carcinogenesis. Science 222:771-778, 1983.

Yunis, J.J.: The chromosomal basis of human neoplasia. Science 221: 227-236, 1983.

IV. Gene Map of Chromosome M (the mitochondrial, or 25th chromosome)

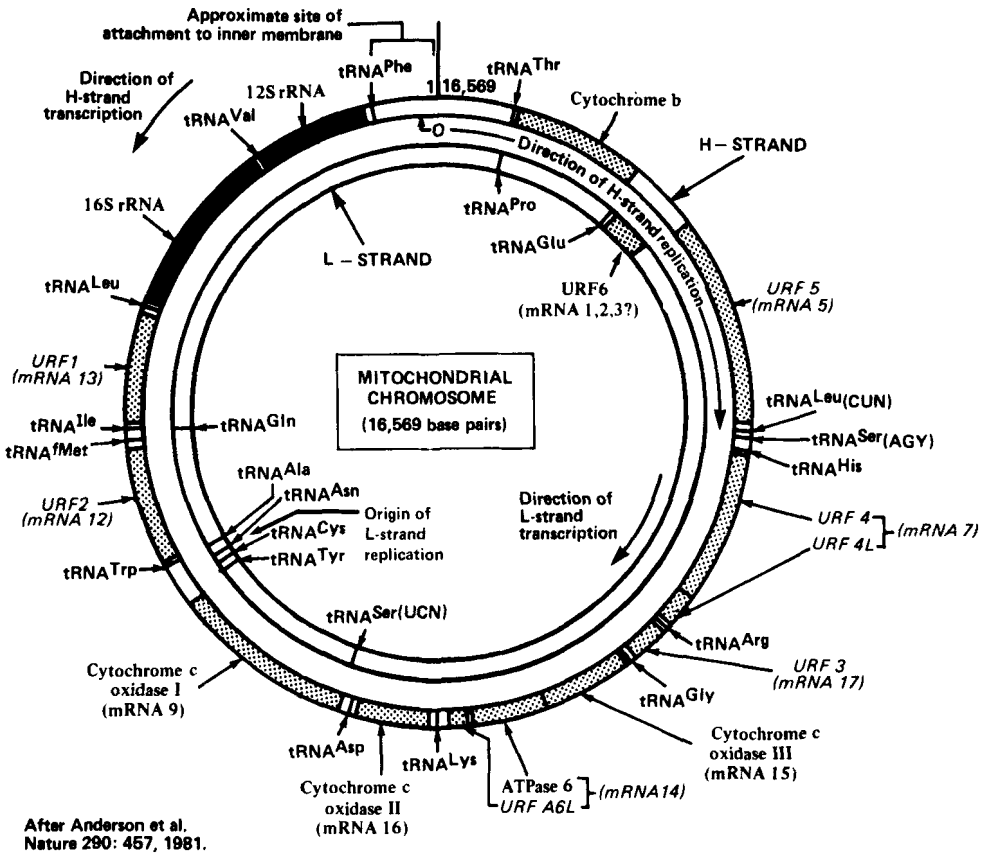
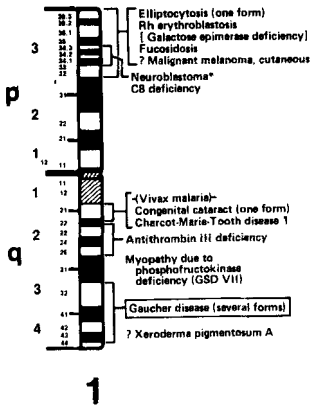
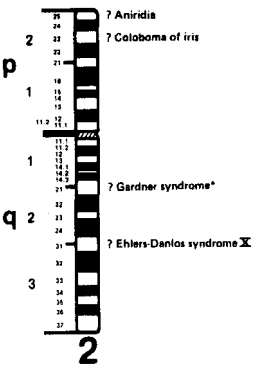
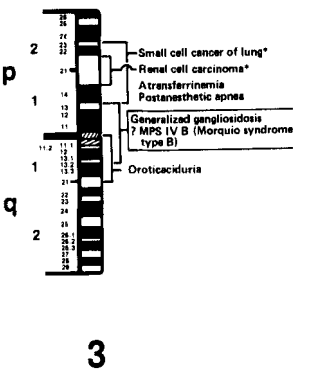


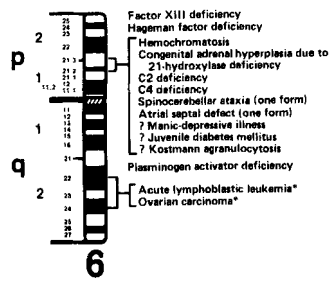
Fig. 11. After Anderson, Bankier, Barrell, de Bruijn, Coulson, Drouin, Eperon, Nierlich, Roe, Sanger, Schreier, Smith, Staden, and Young: Sequence and organization of the human mitochondrial genome. Nature 290:457-465, 1981. The 16,569 base pairs are the equivalent of 5,523 codons. Most of the mitochondrial DNA serves a coding function. The genes contain no intervening sequences, and little in the way of flanking sequences are present. The ribosomal and transfer RNAs are those involved in the synthesis of protein in the mitochondrion. Of the 22 tRNAs, 14 are coded on the L strand. In the case of 7 of the 13 protein coding units, the specific protein determined is identified. The other protein coding units are designated URF (unknown reading frame). URF A6L codes for component 25 (Biochemistry 21: 3188, 1982), and URF1 and URF3 for components 12 and 24, respectively, of the mitochondrial translation products of HeLa; ATPase 6 is component 17 (PNAS 80: 5536, 1983). Only 1 protein is coded by the L strand. See Denaro et al., PNAS 78: 5768, 1981, for description of DNA sequence polymorphism of mtDNA without AA polymorphism. Information on approximate point of attachment to the inner membrane of the mitochondrion from Albring, Griffith, Attardi: PNAS 74:1348, 1977. Cultured human cells resistant to chloramphenicol have a mutation in 16S rRNA (Kearsey and Craig, Nature 290: 607, 1981); Blanc et al., Nucleic Acids Res. 9: 5785, 1981). (Also see map of Attardi, et al., Cytogenet. Cell Genet. 32:85, 1982.) A classic study of the mule and hinny, reciprocal horse-donkey hybrids with horse and donkey as mother, respectively, indicated maternal origin of mtDNA (Hutchison et al., Nature 251:536, 1974). See Giles et al., PNAS 77:6715, 1980 for evidence of maternal inheritance of mtDNA in man. The haploid nuclear genome is about 200,000 times larger than the mitochondrial genome. Anderson et al. filled 3 pages of Nature with the sequence of the mitochondrial genome. To print the nuclear sequence (especially with indications of variations therein) would require a sizeable library. Because of the small size of the ribosomal RNAs, the circular chromosome, chloramphenicol sensitivity of

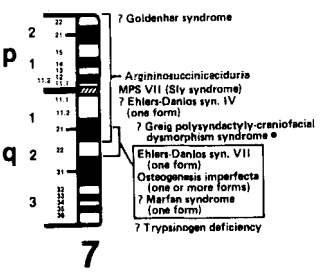
mitochondrial protein synthesis and resistance to choloramphenicol as a mitochondrial mutation, and general morphologic similarities, it has been suggested that the mitochondrial genetic system originated by endosymbiosis of a primitive bacterium. However, the mitochondrial genetic code differs from that of any present day prokaryotes (Barrell et al., Nature 282:189, 1979); e.g., UGA codes for tryptophan (not termination), AUA codes for methionine (not isoleucine), and AGA and AGG code for termination (not arginine). The mitochondrial code is read in a unique fashion. Furthermore, mitochondrial rRNA shows low homology with bacterial rRNA. Thus, although the endosymbiosis hypothesis remains attractive, the endosymbiont may have been quite different from the progenitor of present day prokaryotes (and eukaryotes). Alternatively, selection (which might be quite different for mitochondrial as contrasted with free-living organisms) may have led to radical changes in the captive cellular organelle (D. C. Wallace, Microbiol. Rev. 46: 208, 1982). Mutation in the mitochondrial chromosome has been shown to underlie chloramphenicol resistance in cultured cells (Wallace et al., J. Cell Biol. 67:174, 1975) and has been proposed for the ophthalmoplegia plus syndrome (16510) and Leber optic atrophy, 30890 (Egger and Wilson, NEJM. 309:142, 1983). Differences in the evolution of the mitochondrial and nuclear genomes were reviewed by Birkey (Science 222:468, 1983).

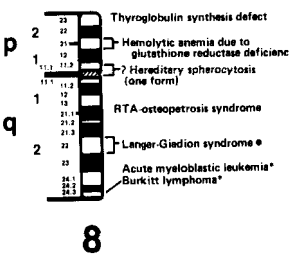

Elliptocytosis (one form)
Rh erythroblastosis
[Galactose epimerase deficiency]
Fucosidosis
? Malignant melanoma, cutaneous
Neuroblastoma*
CB deficiency
-(Vivax malaria)-
Congenital cataract (one form)
Charcot-Marie-Tooth disease 1
Antithrombin III deficiency
Myopathy due to phosphofructokinase deficiency (GSD VII)
Gaucher disease (several forms)
? Xeroderma pigmentosum A
1

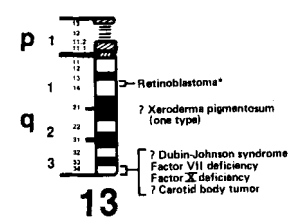

? Aniridia
? Coloboma of iris
? Gardner syndrome*
? Ehlers-Danlos syndrome X
2

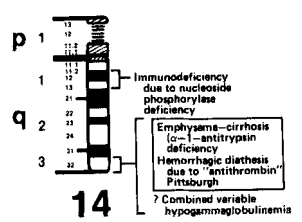

Small cell cancer of lung*
Renal cell carcinoma*
Atransferrinemia
Postanesthetic apnea
Generalized gangliosidosis
? MPS IV B (Morquio syndrome type B)
Oroticaciduria
3

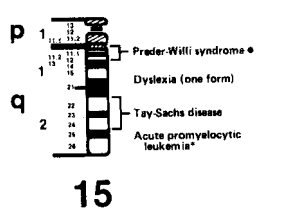

Factor XIII deficiency
Hageman factor deficiency
Hemochromatosis
Congenital adrenal hyperplasia due to 21-hydroxylase deficiency
C2 deficiency
C4 deficiency
Spinocerebellar ataxia (one form)
Atrial septal defect (one form)
? Manic-depressive illness
? Juvenile diabetes mellitus
? Kostmann agranulocytosis
Plasminogen activator deficiency
Acute lymphoblastic leukemia*
Ovarian carcinoma*
6


? Goldenhar syndrome
Argininosuccinicaciduria
MPS VII (Sly syndrome)
? Ehlers-Danlos syn. IV (one form)
? Greig polysyndactyly-craniofacial dysmorphism syndrome
Ehlers-Danlos syn. VII (one form)
Osteogenesis imperfecta (one or more forms)
? Marfan syndrome (one form)
? Trypsinogen deficiency
7


Thyroglobulin synthesis defect
Hemolytic anemia due to glutathione reductase deficienc
? Hereditary spherocytosis (one form)
RTA-osteopetrosis syndrome
Langer-Giedion syndrome
Acute myeloblastic leukemia*
Burkitt lymphoma*
8


Retinoblastoma*
? Xeroderma pigmentosum (one type)
? Dubin-Johnson syndrome
Factor VII deficiency
Factor X deficiency
? Carotid body tumor
13


Immunodeficiency due to nucleoside phosphorylase deficiency
Emphysema–cirrhosis (α–1–antitrypsin deficiency
Hemorrhagic diathesis due to "antithrombin" Pittsburgh
? Combined variable hypogammaglobulinemia
14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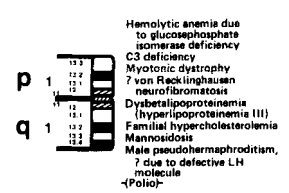
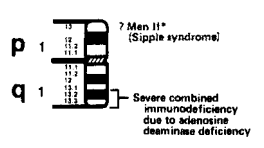
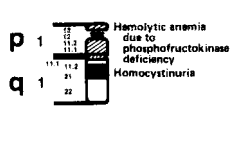
Dyslexia (one form)
Tay-Sachs disease
Acute promyelocytic leukemia*
15


Hemolytic anemia due to glucosephosphate isomerase deficiency
C3 deficiency
Myotonic dystrophy
? von Recklinghausen neurofibromatosis
Dysbetalipoproteinemia (hyperlipoproteinemia III)
Familial hypercholesterolemia
Mannosidosis
Male pseudohermaphroditism, ? due to defective LH molecule
-(Polio)-
19


? Men II*
(Sipple syndrome)
Severe combined immunodeficiency due to adenosine deaminase deficiency
20


Hemolytic anemia due to phosphofructokinase deficiency
Homocystinuria
21

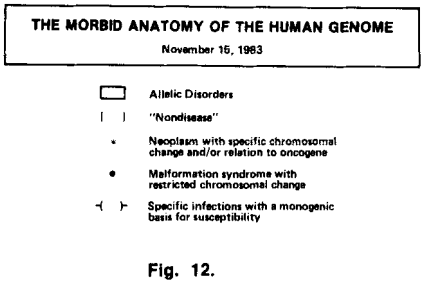
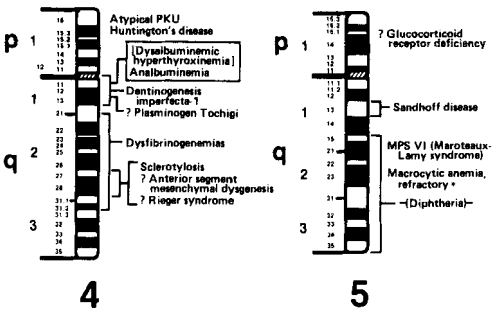
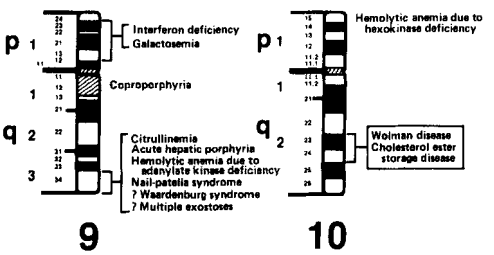
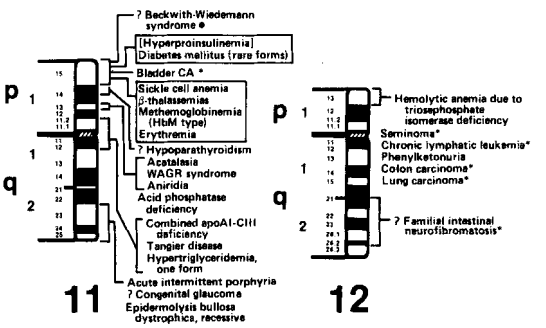
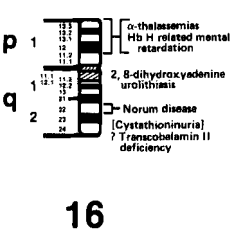

THE MORBID ANATOMY OF THE HUMAN GENOME
November 15, 1983
Allelic Disorders
"Nondisease"
Neoplasm with specific chromosomal change and/or relation to oncogene
Malformation syndrome with restricted chromosomal change
Specific infections with a monogenic basis for susceptibility

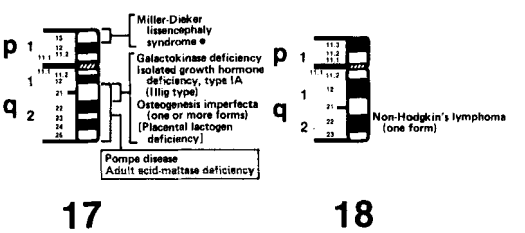
Fig. 12.

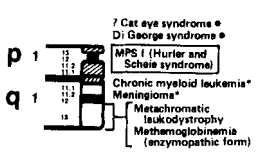

Atypical PKU
Huntington's disease
[Dysalbuminemic hyperthyroxinemia]
Analbuminemia
Dentinogenesis imperfecta-1
? Plasminogen Tochigi
Dysfibrinogenemias
Sclerotylosis
? Anterior segment mesenchymal dysgenesis
? Rieger syndrome
4
? Glucocorticoid receptor deficiency
Sandhoff disease
MPS VI (Maroteaux-Lamy syndrome)
Macrocytic anemia, refractory *
-(Diphtheria)-
5

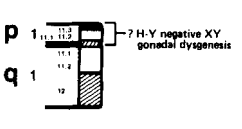

Interferon deficiency
Galactosemia
Coproporphyria
Citrullinemia
Acute hepatic porphyria
Hemolytic anemia due to adenylate kinase deficiency
Nail-patella syndrome
? Waardenburg syndrome
? Multiple exostoses
9
Hemolytic anemia due to hexokinase deficiency
Wolman disease
Cholesterol ester storage disease
10

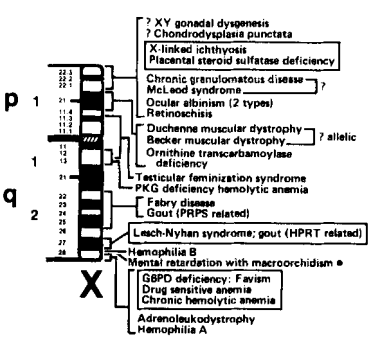

? Beckwith-Wiedemann syndrome •
[Hyperproinsulinemia]
Diabetes mellitus (rare forms)
Bladder CA *
Sickle cell anemia
β-thalassemias
Methemoglobinemia (HbM type)
Erythremia
? Hypoparathyroidism
Acatalasia
WAGR syndrome
Aniridia
Acid phosphatase deficiency
Combined apoAI-CIII deficiency
Tangier disease
Hypertriglyceridemia, one form
Acute intermittent porphyria
? Congenital glaucoma
Epidermolysis bullosa dystrophica, recessive
11
Hemolytic anemia due to triosephosphate isomerase deficiency
Seminoma*
Chronic lymphatic leukemia*
Phenylketonuria
Colon carcinoma*
Lung carcinoma*
? Familial intestinal neurofibromatosis*
12


α-thalassemias
Hb H related mental retardation
2, 8-dihydroxyadenine urolithiasis
Norum disease
[Cystathioninuria]
? Transcobelamin II deficiency
16


Miller-Dieker lissencephaly syndrome •
Galactokinase deficiency
Isolated growth hormone deficiency, type IA (IIIig type)
Osteogenesis imperfecta (one or more forms)
[Placental lactogen deficiency]
Pompe disease
Adult acid-maltase deficiency
17
Non-Hodgkin's lymphoma (one form)
18


? Cat eye syndrome •
Di George syndrome •
MPS I (Hurler and Scheie syndrome)
Chronic myeloid leukemia*
Meningioma*
Metachromatic leukodystrophy
Methemoglobinemia (enzymopathic form)
22


? H-Y negative XY gonadal dysgenesis
Y


? XY gonadal dysgenesis
? Chondrodysplasia punctata
X-linked ichthyosis
Placental steroid sulfatase deficiency
Chronic granulomatous disease
McLeod syndrome
?
Ocular albinism (2 types)
Retinoschisis
Duchenne muscular dystrophy
Becker muscular dystrophy
? allelic
Ornithine transcarbamoylase deficiency
Testicular feminization syndrome
PKG deficiency hemolytic anemia
Fabry disease
Gout (PRPS related)
Lesch-Nyhan syndrome; gout (HPRT related)
Hemophilia B
Mental retardation with macroorchidism •
G6PD deficiency: Favism
Drug sensitive anemia
Chronic hemolytic anemia
Adrenoleukodystrophy
Hemophilia A
X