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Report of the committee on the genetic constitution of chromosomes 7, 8 and 9

M. Smith and M. A. Spence

Chromosome 7

At this conference five new genes and seven random DNA fragments have been assigned to human chromosome 7. The new genes include the beta actin functional gene, ACTB and an actin pseudogene (provisional assignments), the neuropeptide Y gene NPY (provisional assignment), the T cell receptor beta gene (confirmed assignment) and the T cell receptor gamma gene (confirmed assignment). The seven random DNA fragments provisionally assigned to chromosome 7 have all been regionally assigned, and four of these fragments have been shown to detect restriction fragment length polymorphisms. Two genes which were previously assigned to chromosome 7 have been further regionally assigned: the COL1A2 collagen gene to 7q21-7q22, and the (EGFR ERBB) gene to chromosome 7p14-p12. RFLPs have been detected with the COL1A2 genes and linkage analysis between these and various forms of Osteogenesis imperfecta were described. In the literature since HGM7 7 there are reports of one new gene assignment to chromosome 7: an arginosuccinate synthetase pseudogene was assigned to chromosome 7 by Daiger et al. (1984). There are also reports of two regional assignments on chromosome 7: the gene coding for phosphoserine phosphatase was assigned to the region 7pter-q22, by Koch et al., (1984) and the gene coding for biliverdin reductase (BLVR) was assigned to 7pter-q22 by Parkar et al. (1984) overlapping the initial assignment to 1p14-gen (Meera Khan et al. 1983).

A specific probe for a beta actin functional gene which is expressed in fibroblasts was isolated by Ng et al. (HGM8). Using this probe a beta actin functional gene designated ACTB, was assigned to chromosome 7 and specifically to the region 7pter-7q22 through Southern blot hybridization of EcoRI fragments of human, rodent and somatic cell hybrid DNA. The ACTB probe recognized a 14kb fragment in human DNA but did not hybridize with mouse DNA. A probe isolated from the 3' untranslated region of the actin gene was found to detect a series of actin pseudogenes. This probe designated HAS, hybridized to chromosomes 5, 7, 18 and X. The beta actin pseudogene on chromosome 7 is referred to as ACTBP5 and is located in the region 7q22-7qter.

Takeuchi et al. (HGM8) used a cDNA clone for neuropeptide Y (NPY) to examine the chromosomal location of the gene coding for this polypeptide. cDNA clones were derived from human pheochromocytoma tissues. The NPY probe hybridized with 3 PstI fragments of human DNA; these fragments were 6.7, 4.9 and 0.5kb in size. This probe also hybridized with mouse DNA fragments which were 9.2 and 2.6kb in size. In somatic cell hybrid DNA, occurrence of human genomic DNA fragments which hybridized to the NPY probe was dependent on the presence of human chromosome 7, and specifically on the presence of the 7pter to 7q22 region.

Three groups have localized human beta and gamma T cell receptor genes on human chromosome 7. These two genes are expressed in T cells and undergo rearrangement during differentiation. Collins et al. (1984) assigned the T cell receptor beta gene to chromosome 7 and specifically to the region 7q22-qter. Morton et al., (HGM8) used somatic cell hybrid DNA digested with SstI to confirm the assignment of this gene to chromosome 7 q22-qter. In addition they utilized *in situ* hybridization to localize the T cell receptor beta chain gene to band 7q32. Le Beau et al. (1984) reported

localization of the human T cell receptor beta chain gene to the region 7q35 on the basis of *in situ* hybridization studies.

Spurr et al. (HGM8) have derived cDNA probes for the T cell receptor gamma gene by subtraction cloning of a T cell receptor related gene which undergoes rearrangement during T cell differentiation. They report regional localization of this gene to the region 7pter-7q22. Morton et al. (HGM8) derived a genomic clone for the constant region of the T cell gamma chain gene and a cDNA clone for the variable region of this gene. The cDNA clone for the variable chain detected SstI fragments of 5.0 and 4.5kb in human genomic DNA and in rodent human hybrid cell DNA where human chromosome 7 or the 7pter-7q22 region was present. *In situ* hybridization was carried out using the genomic clone for the T cell receptor gamma gene. Based on results of these experiments Morton et al. assign this gene to the 7p15 band.

Bartels et al., (HGM8) have regionally assigned six cloned DNA sequences to human chromosome 7. Four of these sequences map to the region 7cen-7q22; they are referred to as pJu48, pB79a, pJ3.11 and pJu28. The pJu48 sequence detects a TaqI polymorphism while the pJ3.11 probe detects MspI and TaqI polymorphisms. One sequence, pJ5.11, which maps to the 7pter-7p14 region, detects a MspI polymorphism. The pB78 sequence maps to the region 7pter-q22.

Spurr et al. (HGM8) examined chromosomal assignment and restriction fragment length polymorphism of phage clones isolated from a library of EcoRI digested human DNA cloned into the vector Charon 4A. One such phage, designated by them as phage 6, was assigned to chromosome 7, 7pter-7q22.

Tsipouras et al. (1984), (HGM8) have regionally assigned the human procollagen gene COL1A2 to 7q21-q22. The COL1A2 gene has been shown to be polymorphic with a number of different restriction enzymes including: EcoRI (variant allele frequency 0.38), MspI (variant allele frequency 0.14) and StuI (variant allele frequency 0.09). These RFLPs have been shown to be in linkage disequilibrium; no evidence of recombination between the EcoRI and MspI RFLPs have thus far been obtained. Tsipouras et al. (HGM8) demonstrated close linkage between the dominantly inherited type IV Osteogenesis imperfecta and the COL1A2 EcoRI polymorphism: $\theta=0$, $z=3.91$. No evidence was obtained for linkage between COL1A2, EcoRI or MspI polymorphisms and the dominantly inherited type I Osteogenesis imperfecta. These findings therefore imply heterogeneity of linkage between type I and type IV Osteogenesis imperfecta.

Using *in situ*-hybridization Retief et al. (HGM8) have regionally assigned the COL1A2 gene to 7q21.3-7q22.1. The authors further report that when COL1A2 related probes are used in low stringency *in situ* hybridization experiments, significant hybridization to additional chromosomal regions is observed. One such cross hybridizing region (COLL5) is 7p15-pter. Retief et al. propose that additional collagen like genes may be located in these cross hybridizing regions.

Relationship of the pro alpha2I collagen to Marfan's syndrome has been investigated. In one patient an atypical alpha2 collagen chain was found in which the chain was extended by 20 amino acids. (Byers et al. 1981). RFLP's at the collagen gene locus were examined in a large kindred with 21 patients with Marfan's syndrome by Borresen et al. (HGM8). One recombinant was found. The authors suggest that this finding excludes that the genetic abnormality in Marfan's resides within the proalphaI collagen locus in the specific kindred studied by them. It is of interest that the patient described by Byers and patients in the kindred studied by Borresen, differed in their clinical manifestations.

Through analysis of epidermal growth factor receptors in human mouse somatic cell hybrids, Shimizu et al. (1980) and Kondo and Shimizu (1983) assigned the gene coding for these receptors to 7 p13-q22. Shimizu et al. (1985) have utilized DNA probes for EGFR and Southern blot analysis to confirm this assignment. In addition they have demonstrated through Southern blot analysis that DNA probes for the v-erbB genes map to the same region. These authors utilized *in situ* hybridization studies to

demonstrate that both EGFR and v-erb B genes map to the p14-p12 region of chromosome 7. The authors consider this finding as support for the hypothesis that the EGFR gene constitutes a c-erbB protooncogene. Additional support for this hypothesis is the fact that the EGF receptor possesses tyrosine specific protein kinase activity. Shimizu et al. (1985) have sought to determine whether EGFR is involved in malignant transformation. The epidermoid carcinoma cell line A431 has been found to produce a high number of EGF receptors. This cell line was shown to contain a marker chromosome derived from chromosome 7. This marker chromosome carried amplified EGFR genes. In addition the A431 cell line produced a variant EGFR mRNA. This variant mRNA was shown to be the result of an intrachromosomal rearrangement apparently within the EGFR gene.

Yang-Feng et al. (HGM8) have used a DNA probe for EGFR to regionally assign this gene to 7p13.q11.2

Pajunen et al. (HGM8) have utilized monoclonal and polyclonal antibodies to the beta sub-unit of human prolyl 4 hydroxylase to examine production of this enzyme by human rodent somatic cell hybrids. Preliminary studies indicate that the gene coding for the beta sub-unit of prolyl 4 hydroxylase may be located on chromosome 7.

Trent et al. (HGM8) describe isolation and mapping of a presumptive clone for p glycoprotein. This clone was isolated from a colchicine resistant CHO line - CH^RB-30 in which p glycoprotein is amplified. This clone was found to hybridize to human DNA. Studies on somatic cell hybrids indicate that this gene is on human chromosome 7. In situ hybridization studies indicate location on human chromosome 7q36 and hamster chromosome 1q38.

Chromosome 8

At this conference two new genes have been assigned to chromosome 8. These are the gene for tissue type plasminogen activator (confirmed assignment) and the luteinizing hormone releasing hormone gene (provisional assignment). In addition a DNA region homologous to the chicken cardiac myosin light chain gene, has been assigned to chromosome 8. A DNA region corresponding to the junction fragment of a t(8:14)(q24:q11) translocation in a leukemic cell line, has been cloned. A confirmed new regional assignment on chromosome 8 is that of the human thyroglobulin gene to the region 8q23-8q24 by Bernardi et al. (HGM8) and Landegent et al. (HGM8); Berge Le Franc et al., 1985). Since HGM7 there is one report in the literature of a new gene assigned to human chromosome 8: this is the regulatory gene for blood clotting factor 7 (proconvertin), which was assigned to the long arm of chromosome 8 by de Grouchy et al. (1984), on the basis of deletion mapping. Langer-Giedion syndrome (Tricho-rhino-phalangeal syndrome) was further regionally assigned to chromosome 8q22-q24 by Fryns et al. (1983) and Buhler et al. (1984). These latter authors raise the possibility of a second Tricho-rhino-phalangeal syndrome which they assign to the region 8q24.

Benham et al. (HGM8) have isolated clones for human tissue plasminogen activator from a cDNA library made from the Bowes melanoma cell line. Two clones designated ptPA4352 and ptPA-3, have been used to examine EcoR1 fragments of human DNA, rodent DNA and hybrid cell DNA. The presence of human plasminogen activator DNA was shown to be concordant with the presence of human chromosome 8. An EcoR1 RFLP was detected within the plasminogen activator gene. Two EcoR1 fragments occurred: a 2.9kb and a 2.5kb fragment. These fragments were apparently determined by two alleles which each occurred with a frequency of approximately 0.5 in the Caucasian population. Rajput et al. (HGM8) used a cDNA probe for tissue plasminogen activator (65,000 dalton protein) to assign the plasminogen activator gene (PLAT) to human chromosome 8. Visse et al. (HGM8) used a cDNA probe designated ptPA8FL, for the human plasminogen activator gene (PLAT) to assign this gene to human chromosome 8. The ptPA8FL probe for PLAT hybridized to a 6.8kb Pst1 fragment of human DNA and a 3kb fragment of mouse DNA.

Yang-Feng et al. (HGM8) have used a cDNA clone for human tissue type plasminogen activator, and in situ hybridization to regionally assign this gene to human chromosome 8p12.

Yang-Feng et al. (HGM8) assigned a gene encoding the precursor molecule of human luteinizing hormone releasing hormone (LHRH) to human chromosome 8p11.2-p21 using in situ hybridization methods. Southern blot analysis of EcoRI digested human DNA revealed that the LHRH probe hybridized to an 11.5kb fragment. Occurrence of this fragment in Chinese hamster x human somatic cell hybrids was dependent on the presence of human chromosome 8.

McBride et al. (HGM8) assigned the gene for the beta sub-unit of DNA polymerase to chromosome 8. The probe used for this assignment was derived from a library of rat brain cDNA in the lambda gt11 expression vector. The clone was isolated using a polyclonal antibody to chick beta sub-unit of DNA polymerase. The probe was verified by identity of part of the nucleotide sequence with amino acid sequence of oligopeptides from a few regions of the beta sub-unit of DNA polymerase from rat and chick. A 438 base fragment derived from the original clone was used to demonstrate that the gene coding for DNA polymerase beta segregated with chromosome 8.

The carbonic anhydrase alpha2 gene was assigned to chromosome 8 by Venta et al. (1983). Lee et al. (1985) have described RFLPs associated with this gene. A TaqI polymorphism occurs in the 5' flanking region of this gene. Absence or presence of the TaqI restriction site leads to occurrence of a DNA fragment of 5.4kb or fragments of 4.0 and 1.4kb. Each allele occurs with a frequency of 0.5.

Butterworth et al. (HGM8) isolated a cDNA clone for rabbit carbonic anhydrase 1 from a rabbit reticulocyte cDNA library using an oligonucleotide probe. The authenticity of the clone was provided by nucleotide sequencing. This clone hybridized to human DNA and studies in rodent human hybrids indicated that the human carbonic anhydrase 1 gene is on chromosome 8.

Edwards et al. (HGM8) isolated a clone for human carbonic anhydrase III by antibody screening of a human muscle cDNA library in a lambda gt11. This clone was used to determine that the human CA3 gene maps to human chromosome 8.

One sub-family of beta tubulin genes in humans has been found to consist of an expressed gene and three processed pseudogenes (Hall et al. 1983; Lee et al. 1983). One of the pseudogenes in this sub-family referred to as M4ORP maps to human chromosome 8. (Floyd-Smith et al. HGM8)

Balazs et al. (HGM8) have isolated human genomic DNA sequences which are homologous to chicken cardiac myosin light chain cDNA. A clone containing one such fragment was designated PHML 2.1. This clone and the chicken cDNA clone were shown to hybridize to a 9.2kb EcoRI fragment of human DNA. A human genomic clone which contained a 9.2 EcoRI insert and which hybridized to the chicken cardiac myosin clone, was sub-cloned and used for mapping. Positive hybridization of this clone with rodent human hybrid cell DNA, was dependent on the presence of human chromosome 8.

Mathieu-Mahul et al. (HGM8) derived DNA library from a human T cell leukemia line which carried an 8:14 chromosomal translocation, t(8;14) (q24;q11). This library was then screened with a probe derived from Exon 3 of the c-myc gene. One clone, designated K40, contained a 15kb DNA insert which consisted of a 4.5kb insert derived from chromosome 8 (mycEX3) plus a 10.5kb fragment derived from chromosome 14.

Rappold et al. (1984) described three Burkitt lymphoma lines with 2p;8q chromosomal rearrangements, which they examined with probes for c-myc and for Ig kappa chain. They reported that in these translocations the variable portion of the kappa Ig gene remained on 2p- whereas the constant portion of this gene was translocated to 8q. In situ hybridization studies indicate that the c-myc gene is situated at the 5' end of the Ig kappa constant gene in the translocated chromosome.

Bernardi et al. (HGM8) utilized a probe containing exons 9 and 10 of the human thyroglobulin gene for in situ hybridization studies. Results of these studies

indicated that 75 % of labelling occurred in the 8q23-24 region while 25 % of grains occurred in the 8q22-23 region.

Further evidence for assignment of the gene coding for thyroglobulin to band 8q24 was obtained by Landegent et al. (HGM8) who used alpha acetylaminofluorene modified probes with *in situ* and Southern hybridization techniques.

In situ hybridization studies and flow blot analysis on sorted chromosomes were used by Caubet et al. (HGM8) to map the c-mos oncogene to chromosome 8q11. This assignment is inconsistent with the previous assignment of this gene to 8q22 (Neel et al. 1982).

Chromosome 9

At this conference one new gene, two pseudogenes and two arbitrary genomic DNA fragments were assigned to chromosome 9. The new gene assignment is cytoplasmic aldehyde dehydrogenase.

Linkage analysis between alpha (INFA) and beta (INFB) interferon genes is reported. These two genes were previously assigned to the short arm of chromosome 9. Investigations of ABO in families with the breast-ovarian cancer phenotype are reported. A survey of the literature reveals reports of the assignment of seven new genes to chromosome 9. These include genes coding for 2 enzymes, coproporphyrinogen oxidase, methylthioadenosinephosphorylase. In addition the genes coding for two forms of relaxin H1 and H2 were assigned to chromosome 9 by Crawford et al. (1984). Two arginosuccinate synthetase pseudogenes were assigned to chromosome 9: assignment of the ASSP3 gene to the region 9q11-q22 was confirmed, while the ASSPP12 gene was provisionally assigned to chromosome 9q13-q11 Daiger et al. (1984). Regional assignment of the gene coding for galactose-1-phosphate uridyl transferase to 9p13 was confirmed Shih et al. (1984).

Cytoplasmic aldehyde dehydrogenase (ALDH1) was assigned to the long arm of chromosome 9 by Hsu et al. (HGM8) using cDNA probes for this gene and 19 rodent human hybrids.

Two different probes were used. One almost full length cDNA probe (1.579kb) detected six EcoRI fragments in human DNA; one of these fragments was 1.9kb in length. A second probe was a 0.285kb fragment from the cDNA clone. It distinguished 2.9 and 1.9kb fragments in human DNA. Both probes cross hybridized with hamster and mouse DNA. However the 1.9 and 2.9kb bands could be readily distinguished from EcoRI fragments of rodent DNA.

Grandchamp et al. (1983) assayed coproporphyrinogen oxidase in rodent, human and hybrid cell lines using isoelectric focusing followed by quantitative assay of gel fragments for enzyme activity with 3H coproporphyrinogen as substrate. Human coproporphyrinogen focused at pH5.2, the rodent enzyme focused at pH6.3 and in hybrid cell lines positive for human chromosome 9 a biphasic pattern of enzyme activity was observed. Assignment of the gene (CPO) for enzyme which represents the sixth enzyme in the heme biosynthesis pathway, is of interest in view of the previous assignment of aminolevulinic acid dehydratase (ALAD) which represents the second enzyme in the heme biosynthesis pathway.

Methylthioadenosinephosphorylase gene (MTAP) was assigned to chromosome 9 by Carrera et al. (1984), using rodent human somatic cell hybrids and isoelectric focusing. Following electrophoresis enzyme activity was blotted to DE 81 paper which was then reacted with the substrate $3\text{H}_5'$ deoxy $5'$ methylthioadenosine. The product of the enzyme reaction, 3H methylthioribose-1-phosphate adhered to the anion exchange paper and following extensive washing of the paper, could be detected by autoradiography.

To chromosomally assign the structural genes for Relaxin hormones. Crawford et al., (1984) used genomic DNA clones and cDNA clones isolated from a pregnant ovary

cDNA library. Genes coding for H1 and H2 Relaxin, referred to as RLN1 and RLN2 respectively, were regionally assigned to chromosome 9pter-9q12.

Henry et al. (1985) used cDNA probes to map the structural gene for aldolase B. Studies on somatic cell hybrids indicated localization of this gene to chromosome 9. Regional localization was carried out using gene dosage. Results indicated assignment of ALDOB to cen-q32. In situ hybridization studies indicate localization to 9p21.3-9q22.2.

Icking et al. (HGM8) have assigned to chromosome 9p24-pter a probe referred to by them as DR6 (gene name D9S1). This probe is a 3.5kb arbitrary non-repetitive human genomic DNA fragment which detects polymorphic sequences with the enzymes HindIII and SstI.

A random DNA probe cloned in lambda and designated phage 42 (D9S4) by Spurr et al., (1985), has been assigned to chromosome 9. This probe detects polymorphism with BglII, Taq and Msp.

Previous studies (Skolnick et al. (1984) suggested possible linkage between a breast cancer susceptibility locus and the ABO locus on 9q34 (lod. 1.43). In the study reported at this conference by Anderson et al., (HGM8) linkage of ABO and the breast-ovarian cancer phenotype was evaluated in 11 pedigrees with 340 different members. No evidence of linkage with ABO was found. The report of Skolnick et al. of single pedigree did not contain any cases of breast and ovarian cancer and may represent a separate entity.

Allderdice et al., (1986) have analyzed ABO, AK and ORM in 3 Newfoundland kindreds in whom a specific pericentric inversion of chromosome 9 was segregating. Their studies suggest that ABO maps to 9q22.1-q34.1. AK maps to 9q34.1-q34.3, while the ORM gene maps to the region 9q34.3-qter. No additional lod scores were submitted to HGM8, therefore no further information on the order in this linkage group was available. The lod scores remain as summarized in Fig. 4 of HGM7 and the compilation by Keats (1981).

Ohlsson et al. (HGM8) have carried out linkage analysis on interferon genes. The alpha (IFNA) and beta (IFNB) interferon gene families are located on chromosome 9 p. Variability within these interferon gene families is very high so that these probes constitute valuable genetic markers. No recombinants between the alpha and beta genes could be demonstrated in 25 families suggesting that these genes may cluster within several hundred kb.

Table I. Regional assignments to chromosome 7

Regional assignment	Gene symbol	Name or probe	Mode	Status	References
pter-p15	COLL5	Collagen-like 5	A	P	Retief et al. (HGM8)
pter-p14	GCTG	Gamma-glutamyl-cyclotransferase	S	P	Bissbort et al. (1984)
pter-p14	D7S10	pJ5.11	S	P	Bartels et al. (HGM8)
pter-q22	D7S11	phage 6	S	P	Spurr et al. (HGM8)
pter-q22	NPY	Neuropeptide Y	S,RE	P	Takeuchi et al. (HGM8)
pter-q22	ACTB	Actin, beta	S,RE	P	Ng et al. (HGM8)
pter-q22	PSP	Phosphoserine phosphatase	S	C	Koch et al. (1983)
pter-q22	D7S12	pB78	S,RE	C	Cooper et al. (1985); Bartels et al. (HGM8)
p21-q22	ASL	Argininosuccinate lyase	S	P	Naylor et al. (1978)
p15	ASSP11	Argininosuccinate synthetase pseudogene 1 pA55pc7	S	P	Beaudet et al. (HGM8); Daiger et al. (1984)
p15	TCRG	T cell receptor (rearranging), gamma polypeptide	S,RE	C	Spurr et al. (HGM8); Morton, Duby et al. (HGM8)
p14-cen	BLVR	Biliverdin reductase	S	C	Meera Khan et al. (1983); Parkar et al. (1984)
p13-p11	EGFR	Epidermal growth factor receptor	S	C	Robson and Meera Khan (HGM5)
p13-q22	MDH2	Malate dehydrogenase, NAD (mitochondrial)	S	C	Bootsma and Ruddle (HGM4)
p12-p14	ERBB	Avian erythroblastic leukemia viral (v-erb-b) oncogene homolog	S,RE	C	Spurr et al. (1984); Sakaguchi et al. (1984); Yang-Feng, Schechter et al. (HGM8); Zabel et al. (1984)
p11.2	FRA7A	Fragile site, rare folic acid type, fra(7)(p11.2)	CH	C	Pavey and Webb (1982)
p11-q11	ASNS	Asparagine synthetase	S	P	Arfin et al. (1983)

Table I. Gene assignments to chromosome 7 continued

Regional assignment	Gene symbol	Name or probe	Mode	Status	References
cen-q11.2	MYH5	Myosin heavy polypeptide, skeletal muscle, 5 adult	A	P	Barton et al. (HGM7)
cen-q22	D7S6	pJu48	S	P	Bartels et al. (HGM8)
cen-q22	D7S8	pJ3.11	S	P	Bartels et al. (HGM8)
cen-q22	GUSB	Glucuronidase, beta	S,RE	C	Bootsma and Ruddle (HGM4)
cen-q22	D7S13	pB79a			Cooper et al. (1985); Bartels et al. (HGM8)
cen-q22	D7S14	pJu28			Cooper et al. (1985); Bartels et al. (HGM8)
q21.3- q22.1	OI4	Osteogenesis imperfecta type IV	F	P	Tsipouras et al. (HGM8)
q21.3- q22.1	COL1A2	Collagen, type I, alpha 2 NJ3, NJ1, 12016 HPC1, genomic 3'	S,RE S RE S RE	C	Robson and Meera Khan (HGM6); Grobler-Rabie et al. (1985); Brebner et al. (1985); Retief et al. (HGM8)
q22 or q32-36	H1	H1 histone	A	C	Robson and Meera Khan (HGM6)
q22 or q32-36	H2A	H2A histone	A	C	
q22 or q32-36	H2B	H2B histone	A	C	Robson and Meera Khan (HGM6)
q22 or q32-36	H3F1	H3 histone family 1	A	C	Robson and Meera Khan (HGM6)
q22 or q32-36	H4F1	H4 histone family 1	A	C	Robson and Meera Khan (HGM6)
q22-qter	ACTBP5	Actin, beta pseudogene 5	S,RE	P	Ng et al. (HGM8)
q22-qter	NM	Neutrophil migration	D	P	de la Chapelle et al. (HGM6)
q22-qter	TRY1	Trypsin 1	S,RE	P	Honey et al. (HGM7)

Table I. Gene assignments to chromosome 7 continued

Regional assignment	Gene symbol	Name or probe	Mode	Status	References
q32 or q35	TCRB	T cell receptor (rearranging) beta polypeptide B671	S	C	Barker et al. (1984); Collins et al. (1984); Morton, Doby et al. (HGM8); Le Beau et al. (1985)
q36	PGY1	P glycoprotein 1 lambda CHP1	S,RE	P	Trent et al. (HGM8)
	CPA	Carboxypeptidase A	S,RE	P	Honey et al. (HGM7)
	D7S1	pA2H3	S,RE	P	Humphries et al. (1983)
	D7S2	p	S,RE	P	Balazs et al. (HGM7)
	D7S3	p7-11	S,RE	P	Balazs et al. (HGM7)
	D7S4	p7-12	S,RE	P	Balazs et al. (HGM7)
	D7S5	p7-13	S,RE	P	Balazs et al. (HGM7)
	GCF1	Growth control factor 1	S	P	Donald et al. (1982)
	HADH	Hydroxyacyl-CoA dehydrogenase	S	P	Craig et al. (HGM3)
	NHCP2	Non-histone chromosome protein 2 NHCP2	S	P	Alevy and Fleischman (1980)
	P04DB	Proline, 2-oxoglutarate dioxygenase, beta polypeptide	S	T	Pajunen et al. (HGM8)
	PYHG3	Protein spot in 2-D gels (MW 106K)	S	P	Taggart and Francke (HGM6)
	PYHG8	Protein spot in 2-D gels (MW 80K)	S	P	Taggart and Francke (HGM6)
	S7	Surface antigen (chromosome 7)	S	P	Cicurel and Croce (1977)
	UP	Uridine phosphorylase	S	C	Robson and Meera Khan (HGM5)

Table II. Gene assignments to chromosome 8

Regional assignment	Gene symbol	Name or probe	Mode	Status	References
p21.1	GSR	Glutathione reductase	S,D	C	Bootsma and Ruddle (HGM5)
p1	SPH1	Spherocytosis	F	C	Kimberling et al. (1975); Bass et al. (1983)
p12	PLAT	Plasminogen activator, tissue ptPA-3 pTR5, ptPA8FL, pW349F	S,RE	C	Benham et al. (HGM8); Rajput et al. (HGM8); Visse et al. (HGM8); Yang-Feng, Opdenakker et al. (HGM8)
p11.2-p21	LHRH	Luteinizing-hormone releasing hormone precursor	S,A	P	Yang-Feng, Seeburg et al. (HGM8)
q11 or q22	MOS	Moloney murine sarcoma viral (v-mos) oncogene homolog M5-2A	RE,A	C	Prakash et al. (1982); Neel et al. (1982); Sakaguchi et al. (HGM7); Caubet et al. (HGM8)
q13-qter	GPT	Glutamic-pyruvate transaminase	S	I	Robson and Meera Khan (HGM6)
q21.1-qter	GLYB	Glycine B complementing	S	P	Kao et al. (HGM7)
q22.3	FRA8A	Fragile site, rare folic acid type fra(8)(q22.3)	CH	C	Meera Khan and Smith (HGM7)
q22-q24	LGS	Langer-Giedion syndrome	D	C	Meera Khan and Smith (HGM7); Buhler et al. (1984); Fryns et al. (1984)
q24	MYC	Avian myelocytomatosis viral (v-myc) oncogene homolog	RE,A	C	Taube et al. (1982); Dalla Favera et al. (1982); Neel et al. (1982); Sakaguchi et al. (1984)
q24	TG	Thyroglobulin pCHT16/8 pCHT16/3.7 pHT0.98	S,RE	C	van Ommen et al. (HGM7); Berge-Lefranc et al. (1985); Landegent (HGM8); Bernardi (HGM8)

Table II. Gene assignments to chromosome 8 continued

Regional assignment	Gene symbol	Name or probe	Mode	Status	References
q24	POLB	Polymerase (DNA), beta polypeptide	A,RE	P	McBride, Merry, Znudka et al. (HGM8)
q24.1	FRA8C	Fragile site, common aphidicolin type, type, fra(8)(q24.1)	CH	P	Yunis and Soreng (1984)
q24.3	FRA8D	Fragile site, common aphidicolin type, fra(8)(q24.3)	CH	P	Yunis and Soreng (1984)
	CA1	Carbonic anhydrase I RCA1	S,RE	P	Butterworth et al. (HGM8)
	CA2	Carbonic anhydrase II H25-3.8	S,RE	P	Venta et al. (1983)
	CA3	Carbonic anhydrase III, muscle specific pCA15	S,RE	P	Edwards et al. (HGM8)
	D14S7	K40	RE	P	Mathieu-Mahul et al. (HGM8)
	F7R	Coagulation factor VII regulator	D	P	DeGrouchy et al. (1984)
	FNZ	Fibronectin, influences presence on cell surface	S	C	Owerbach et al. (1978); Rennard et al. (1981)
	MYLL1	Myosin light polypeptide, cardiac-like HML-2.1	S,RE	P	Balazs et al. (HGM8)
	TUBBP1	Tubulin, beta poly- peptide pseudogene 1 M40	S,RE	P	Floyd-Smith et al. (HGM8)

Table III. Gene assignments to chromosome 9

Regional assignment	Gene symbol	Name or probe	Mode	Status	References
pter-p24	D9S3	DR6	S	P	Icking et al.(HGM8)
pter-p13	IFNA	Interferon, alpha (leukocyte) ple IFA	S,RE	C	Owerbach et al. (1981); Trent et al. (1982)
pter-q11	D9S1	p12-8	S,RE	P	Naylor et al. (1984)
pter-q12	RLN1	Relaxin 1 (H1)	S	P	Crawford et al. (1984)
pter-q12	RLN2	Relaxin 2 (H2)	S	P	Crawford et al. (1984)
p34-qter	ASS	Argininosuccinate synthetase	S	C	Carritt et al. (1977) Daiger et al. (1982) Freytag et al. (1982)
p24-p13	IFNB	Interferon, beta (fibroblast) pFIF3	S,RE	C	Robson and Meera Khan (HGM6)
p24-p13	AK3	Adenylate kinase 3	S	C	Cook et al. (1978)
p22-q32	AC01	Aconitase 1, soluble	S	C	Cook et al. (1978)
p21.3-q22.2	ALDOB	Aldolase B		P	Henry et al. (1985)
p21.1	FRA9A	Fragile site, rare folic acid type (fra(9)(p21.1))	CH	P	Meera Khan and Smith (HGM7)
p13	GALT	Galactose-1-phosphate uridylyltransferase	S,D	C	Meera Khan and Robson (HGM5)
p13-q11	ASSP12	Argininosuccinate synthetase pseudo- gene 12	S,RE	P	Beudet et al. (1982); Daiger et al. (1984)
cen-q34	FPGS	Folylpolyglutamate synthetase	S	P	Jones and Kao (HGM5, HGM7)
cen-qter	GRP78	Glucose regulated protein (MW 78K)	S,RE	P	Law et al. (1984)
q11-q22	ASSP3	Argininosuccinate synthetase pseudo- gene 3 pAS1	S	P	Beudet et al. (1982); Daiger et al. (1984)

Table III. Gene assignments to chromosome 9 continued

Regional assignment	Gene symbol	Name or probe	Mode	Status	References
q12	DNCM	DNA associated with cytoplasmic membrane	A	P	Kuo et al. (1974)
q21-q32	ASS	Argininosuccinate synthetase	S	C	Carritt et al. (1977)
q32	FRA9B	Fragile site, rare folic acid type, fra(9)(q32)	CH	P	Meera Khan and Smith (HGM7)
q32	FRA9E	Fragile site, common aphidicolin type, fra(9)(q32)	CH	C	Glover et al. (1984)
q34	ABL	Abelson murine leukemia viral (v-abl) oncogene homolog ab1K2	S,RE	C	Heisterkamp et al. (1982) De Klein et al. (1982) Bartrom et al. (1983)
q34	AK1	Adenylate kinase 1	S,D	C	Cook et al. (1978)
q34	ABO	ABO blood group	F	C	Cook et al. (1978)
q34	NPS1	Nail patella syndrome 1	F	C	Cook et al. (1978)
q34	WS1	Waardenburg syndrome, type 1	F	T	Cook et al. (1978)
q	ALDH1	Alcohol dehydrogenase (class I) alpha polypeptide	S,RE	P	Hsu et al. (HGM8);
q	ORM	Orosomucoid	F	C	Eiberg et al. (HGM6); Muncie et al. (1979); Falk et al. (1975); Lewis et al. (HGM7); Cox and Francke (1985)
	ALAD	Delta-amino-levulinate dehydratase	F	C	Eiberg et al. (HGM7); Amorim et al. (1979); Beaumont et al. (1984);

Table III. Gene assignments to chromosome 9 continued

Regional assignment	Gene symbol	Name or probe	Mode	Status	References
	CP0	Coproporphyrinogen oxidase	S	P	Grandchamp et al. (1983)
	D9S2	9ER1	S,RE	P	Kao et al. (HGM7)
	D9S4	Phage 42			Spurr et al. (HGM8)
	IGHEP2	Immunoglobulin epsilon polypeptide pseudogene 2	S,RE	P	Batthey et al. (1982)
	MTAP	Methylthioadenosine phosphorylase	S	P	Carrera et al. (1984)
	PYHG13	Protein spot in 2-D gels (MW 40K)	S	P	Taggart and Francke (HGM6)
	PYHG14	Protein spot in 2-D gels (MW 37K)	S	P	Taggart and Francke (HGM6)
	PYHG15	Protein spot in 2-D gels (MW 35K)	S	P	Taggart and Francke (HGM6)
	PYHG16	Protein spot in 2-D gels (MW 38K)	S	P	Taggart and Francke (HGM6)

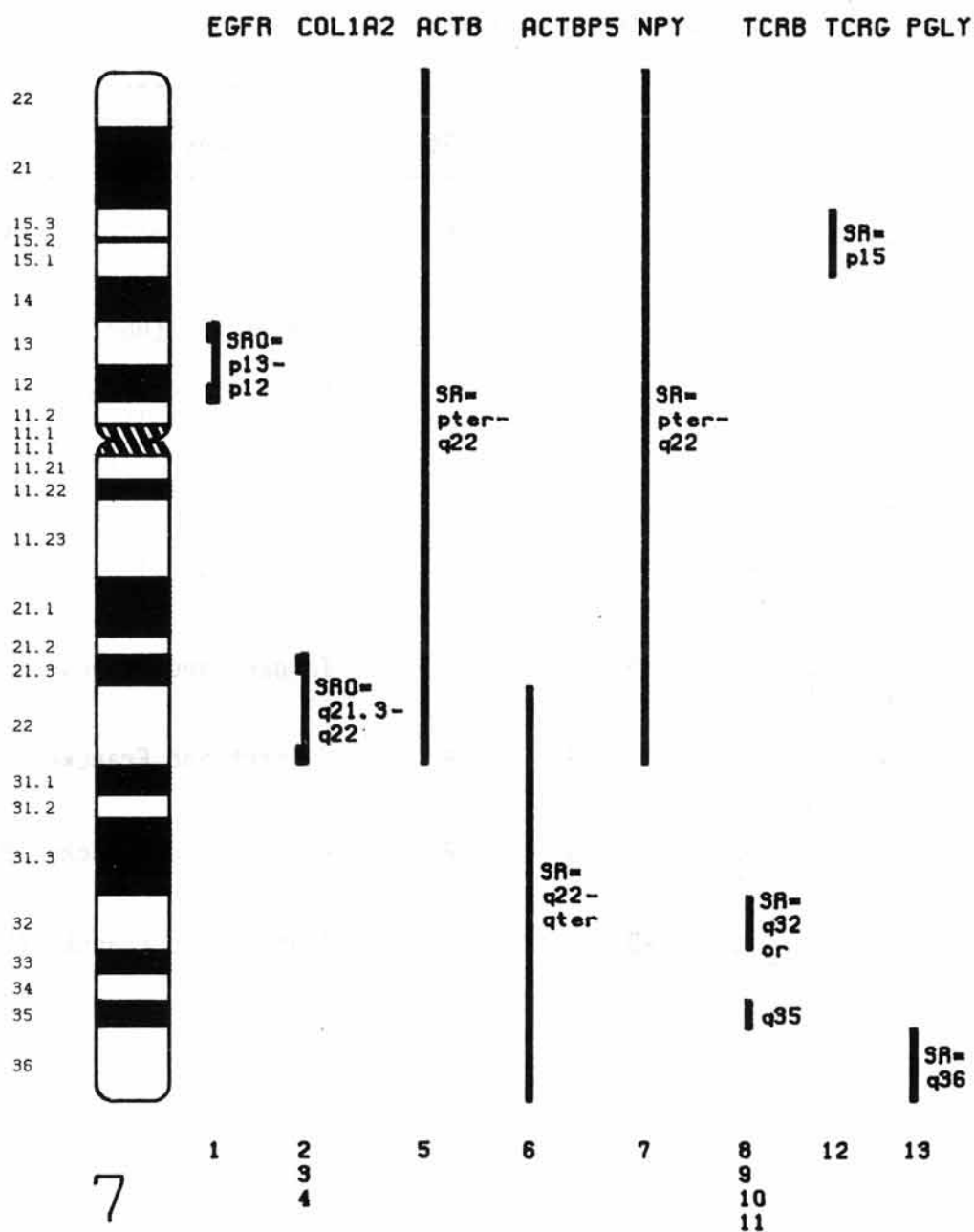


Fig. 1. Regional assignments to chromosome 7. See page 92 for explanations.

References: (1) Shimizu et al. (HGM8); (2) Grobler-Rabie et al. (1985); (3) Brebner et al. (1985); (4) Rettig et al. (1985); (5,6) Takeuchi et al. (HGM8); (7) Ng et al. (HGM8); (8) Collins et al. (1984); (9) Barker et al. (1984); (10) Morton Duby et al. (HGM8); (11) Le Beau et al. (1985); (12) Morton, Duby et al. (HGM8); (13) Trent et al. (HGM8)

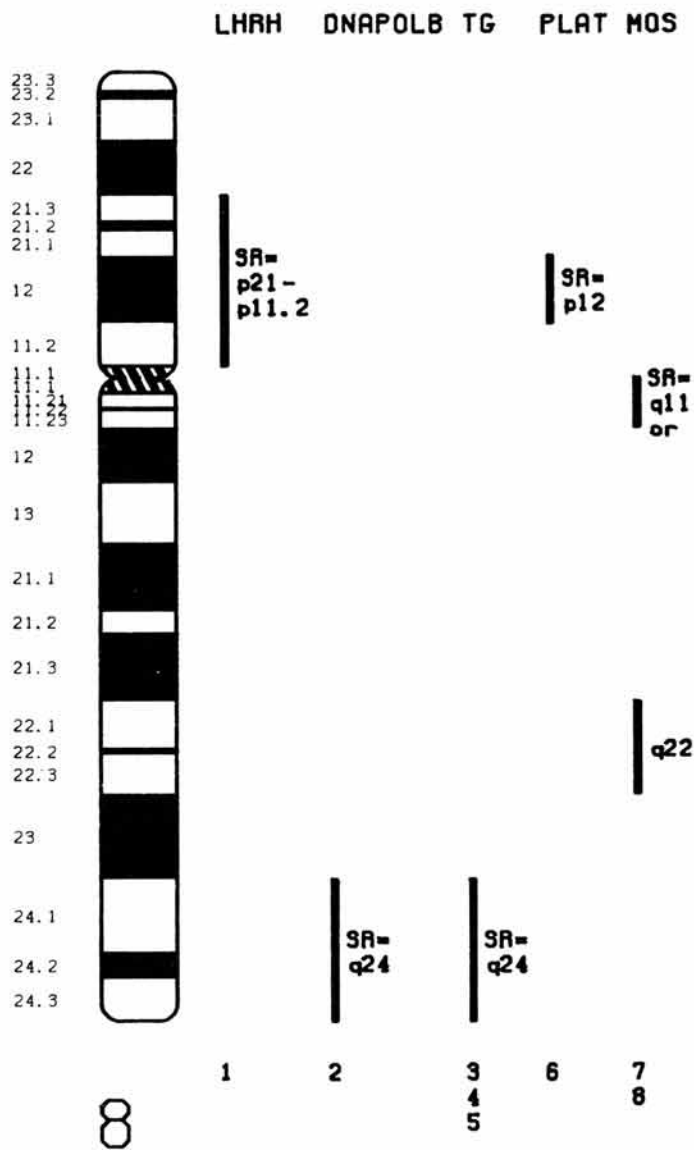


Fig. 2. Regional assignments to chromosome 8. See page 92 for explanations.

References: (1) Yang-Feng, Seeburg et al. (HGM8); (2) McBride, Merry et al. (HGM8); (3) Bernardi et al. (HGM8); (4) Landegent et al. (HGM8); (5) Berge le Franc (1985); (6) Yang-Feng, Opdenakker et al. (HGM8); (7) Caubet et al. (HGM8); (8) Neel et al. (1982)

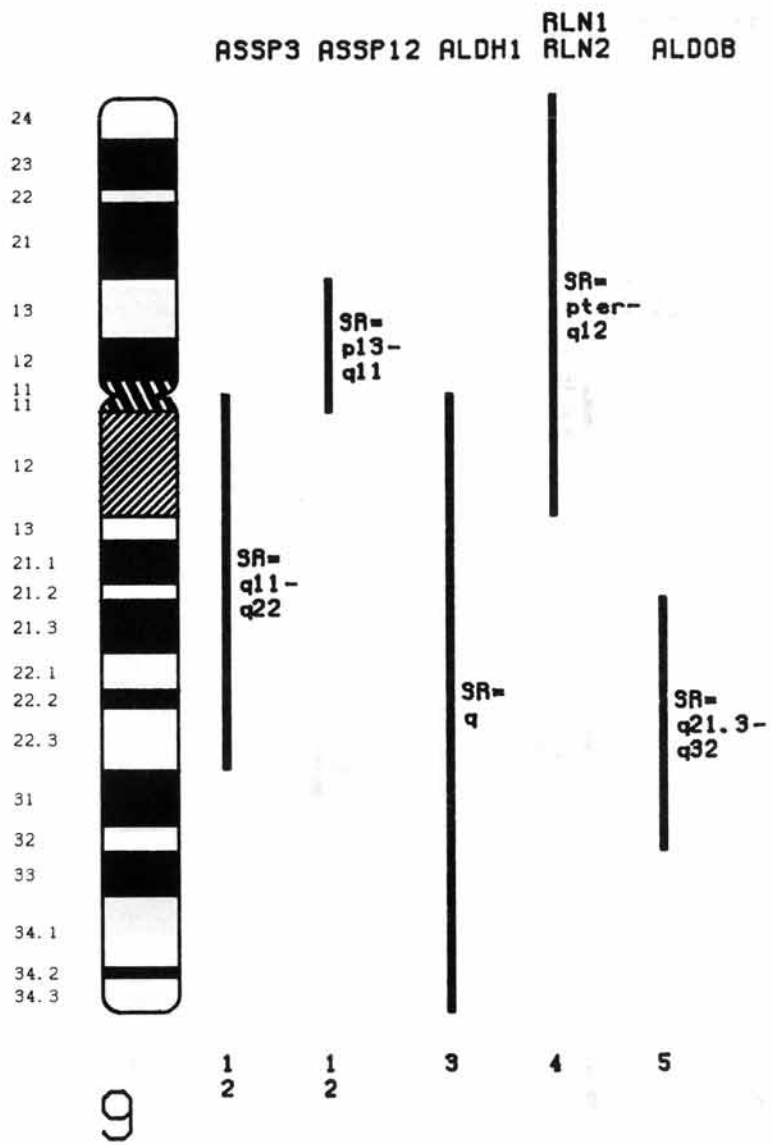


Fig. 3. Regional assignments to chromosome 9. See page 92 for explanations.

References: (1) Beudet et al. (1982); (2) Daiger et al. (1984); (3) Hsu et al. (HGM8); (4) Crawford et al. (1984); (5) Henry et al. (1985);

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