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# Gene mapping and medical genetics

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## Genetic markers on chromosome 7

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**SUMMARY** Chromosome 7 is frequently associated with chromosome aberrations, rearrangements, and deletions. It also contains many important genes, gene families, and disease loci. This brief review attempts to summarise these and other interesting aspects of chromosome 7. With the rapid accumulation of cloned genes and polymorphic DNA fragments, this chromosome has become an excellent substrate for molecular genetic studies.

Chromosome 7 has been a subject of interest in medical genetics because of its frequent association with chromosome aberrations, rearrangements, and deletions, and because of the localisation of several important genes, gene families, and disease loci on this chromosome. It accounts for approximately 5% of the human genome<sup>1</sup> and it is estimated to be 136 centimorgans (cM) in length<sup>2</sup> and to contain 150 million basepairs of DNA. The short (p) and long (q) arm size ratio is approximately 1 to 1.5 (figure), but it varies significantly between prometaphase and midmetaphase.<sup>3</sup>

Approximately 40 genetic loci were reported to be on chromosome 7 at the Eighth Human Gene Mapping Workshop (Helsinki, September 1985).<sup>4,5</sup> There has been a dramatic increase of markers on this chromosome since then. The total number of gene loci and DNA markers characterised now well exceeds 400<sup>6</sup> (figure, table, K-H Grzeschik, 1987, personal communication, L-C Tsui *et al*, unpublished data). In addition, a chromosome 7 specific genomic library has been constructed by the Los Alamos and Lawrence Livermore National Laboratories using flow sorted chromosome 7 from a hamster-human hybrid cell line.<sup>7</sup>

### Genes and gene families

More than 35 genes of assigned functions have been localised to chromosome 7. They are listed in the table. Some of the genes, such as  $\beta$ -actin<sup>1,3</sup> and pro $\alpha$ 2(I) collagen,<sup>39–43</sup> have been cloned and the

gene structure analysed in detail; some have only been assigned by enzyme activities, such as biliverdin reductase<sup>22</sup> and paraoxanase.<sup>51–54</sup>

There are several gene families identified on chromosome 7. The T cell receptor  $\beta$  and  $\gamma$  chain genes have received most of the attention in the past few years. Both genes have complex gene structures and belong to the large immunoglobulin multigene family.<sup>94,95</sup> Expression of these genes requires proper chromosome rearrangement joining different DNA segments together during T cell differentiation.<sup>96</sup> The  $\gamma$  chain gene has been localised to the short arm, region p15.<sup>20,21</sup> The  $\beta$  chain gene has been mapped to 7q3 but different sub-region localisation data have been obtained by different investigators; it is still unclear whether the gene maps to q32<sup>68</sup> or q35.<sup>67,69</sup>

The multidrug resistant phenotype of tumour cells is correlated with increased expression and amplification of the PGY-1 (*mdr-1*) locus which encodes a membrane glycoprotein called P glycoprotein.<sup>97,98</sup> This protein is thought to function as an energy dependent export pump to reduce intracellular levels of anticancer drugs.<sup>99</sup> Molecular cloning and mapping studies showed that the PGY-1 locus probably contains a number of related genes clustered within 7q21→q22.<sup>45,46</sup>

The  $\beta$ -actin<sup>1,3</sup> and the pro $\alpha$ 2(I) collagen<sup>39–43</sup> genes also belong to multigene families but their related members are dispersed elsewhere in the genome.

### Disease loci

A number of disease loci have been mapped to chromosome 7. The metabolic defects argininosucci-

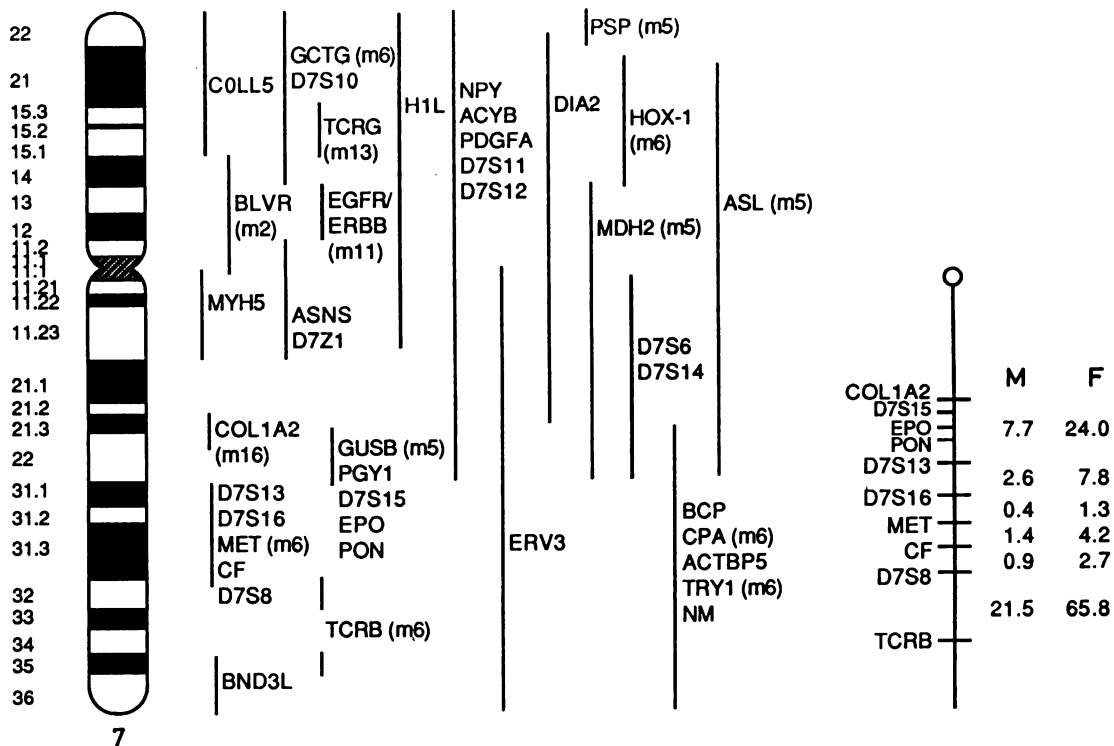


FIGURE Genetic markers on chromosome 7. Regional localisations are indicated by vertical bars. The homologous mouse chromosomes are shown in brackets. The male and female genetic linkage maps for selected loci are shown on the right with numbers in centimorgans.

nicaciduria, mucopolysaccharidosis VII, and trypsinogen deficiency are probably the result of mutations within the genes encoding the enzymes that are involved in the biochemical pathway, namely argininosuccinic lyase,  $\beta$ -glucuronidase, and trypsinogen, respectively.<sup>100</sup> Tritan colour blindness probably results from mutation of the blue-opsin gene.<sup>57</sup>

Osteogenesis imperfecta describes a group of fragile bone disorders which are clinically heterogeneous.<sup>101</sup> The syndrome can be divided into four phenotypic groups depending on the presence or absence of short stature, joint laxity, easy bruising, blue sclerae, presenile hearing loss, and dentinogenesis imperfecta. All of these symptoms are presumed to be the result of mutations in one of the six collagen genes. The genetics of OI have recently been reviewed in this Journal.<sup>102</sup> The pro $\alpha$ 2(I) collagen gene which is involved in all four groups is located on the long arm of chromosome 7 (7q21.3 $\rightarrow$ q22.1).

The collagen genes have also been prime candidates for the gene mutation in Marfan syndrome,

which is a group of autosomal dominant connective tissue disorders with skeletal, cardiac, and ocular defects.<sup>103</sup> A 20 amino acid insertion was found in 50% of the pro $\alpha$ 2(I) chains in one patient and was thought to be the cause of the disease.<sup>104</sup> DNA sequence analysis of the pro $\alpha$ 2(I) collagen gene revealed a 38 bp insertion in this patient.<sup>105</sup> However, subsequent studies showed that this 38 bp insertion is a common DNA fragment length polymorphism among random normal subjects<sup>106</sup> and there is no linkage relationship between the disease locus and the pro $\alpha$ 2(I) collagen gene in family studies.<sup>107 108</sup>

Ehlers-Danlos syndrome is another heterogeneous group of disorders with collagen defects.<sup>100</sup> Their clinical manifestations include laxity of joints, soft and extensible skin which may also be fragile, and excessive bruising. At least one form of this syndrome has been proposed to result from abnormal cleavage of the N-peptide of the pro $\alpha$ 2(I) chain.<sup>109</sup>

Cystic fibrosis (CF) is the most common severe

TABLE *Genes and DNA probes.*

Regional assignment	Gene symbol	Name or probe	Mode* RFLP	References
<i>Genes</i>				
pter→p15	COLL5	Collagen-like 5	A	8
pter→p14	GCTG	γ-glutamylcyclotransferase	S	9
pter→p22	PSP	Phosphoserine phosphatase	S	10
pter→q11	H1L	Histone H1-like genes	A	11
pter→q22	NPY	Neuropeptide Y	S.RE	12
pter→q22	ACTB	β-actin	S.RE	13
pter→q22	PDGFA	Platelet derived growth factor, A chain	RE.S	14
p22-1→q21	DIA2	Diaphorase	S	15, 16
p21→p14	HOX-1	Mouse Hox-1 homologue	RE.S	17
p21→q22	ASL	Arginosuccinate lyase	S.RE	18, 19
p15	TCRG	T cell receptor, γ chain	S.RE +	20, 21
p14→cen	BLVR	Biliverdin reductase	S.RE	22, 23
p13→p11	EGFR	Epidermal growth factor receptor (=ERBB)	S	24-28
p12→p14	ERBB	<i>v-erbB</i> oncogene homologue (avian erythroblastic leukaemia virus)	S.RE	29-32
p13→q22	MDH2	NAD malate dehydrogenase (mitochondrial)	S	33
p11→q11	ASNS	Asparagine synthetase	S.RE +	34, 35
cen→q11.2	MYH5	Myosin heavy polypeptide, adult skeletal muscle	A	36
q21→q22	ERV3	<i>v-erv3</i> homologue	S.RE	37. Tsui <i>et al.</i> , unpublished data
q21.3→q22.1	COL1A2	Collagen, type I α2	S.RE +	38-43
q22	PGY1	P glycoprotein	S.RE,A	44-46
q22	GUSB	β-glucuronidase	S.RE	33, 45, 47, Tsui <i>et al.</i> , unpublished data
q22		G protein, αH subunit	S.RE	FS Collins, J Seidman, 1987, personal communication
q22	EPO	Erythropoietin	S.RE +	48-50
q22	PON	Paraoxonase	F	51-54
q22→q31	MET	met proto-oncogene	RE.A.S +	45, 55, 56
q22→qter	BCP	Blue cone pigment	RE.S	57
q22→qter	CPA	Carboxypeptidase A	RE	58
q22→qter	ACTBP5	β-actin pseudogene 5	S.RE	13
q22→qter	TRY1	Trypsin 1	S.RE +	58, 59
q31	CF	Cystic fibrosis	F	53, 60-63
q32 or q35	TCRB	T cell receptor, B chain	RE.S,A +	64-69
q35→q36	BND3L	Band 3-like, fibroblast	RE.S +	70
	ASSP11	Arginosuccinate synthetase pseudogene 1	RE.S +	71, 72
	PAI-1	Plasminogen activator inhibitor	S.RE	73
		Cytochrome P450 protein glucocorticoid inducible	S.RE +	74
	INFB2	β2 interferon	S.RE +	75
	UP	Uridine phosphorylase	S	76
	HADH	Hydroacyl-CoA dehydrogenase	S	77
	PO4DB	Procollagen-proline, 2-oxoglutarate: 4-dioxygenase, β chain	S	78
<i>DNA segments</i>				
pter→p14	<i>D7S10</i>	pJ5-11	RE.S +	79
pter→q22	<i>D7S11</i>	Phage 6	RE.S +	20
pter→q22	<i>D7S12</i>	pB78	RE.S	79,80
p12→qter	<i>DNF9</i>	pA-8		81
p11→q11	<i>D7Z1</i>	Chromosome 7 specific alpha satellite sequences	RE.S +	82, 83
q22→q31	<i>D7S8</i>	pJ3-11	RE.S +	45, 79
cen→q22	<i>D7S6</i>	pJu48	RE.S +	79
cen→q22	<i>D7S14</i>	pJu28	RE.S	79, 80
q22	<i>D7S15</i>	Lam4-917	RE.S +	45, 60
q22	<i>D7S13</i>	B79a	RE.S,A +	79, 84, V Buckle, 1987, personal communication
q22→q31	<i>D7S16</i>	7C22	RE.S,A +	85, 86
	<i>D7S1</i>	pA2H3	RE.S +	87, 88
	<i>D7S2</i>	p7-15	RE.S	81
	<i>D7S3</i>	p7-11	RE.S	81
	<i>D7S4</i>	p7-12	RE.S	81
	<i>D7S5</i>	p7-13	RE.S	81

(Continued)

TABLE Genes and DNA probes (continued).

Regional assignment	Gene symbol	Name or probe	Mode* RFLP	References
Other determinants q22→qter	NM	Neutrophil migration	D	89
	GCF1	Growth control factor 1	S	90
	NHCP2	Non-histone chromosome protein 2	S	91
	PYHG3	Protein spot in 2D gel MW 106 000	S	92
	YHG3	Protein spot in 2D gel MW 80 000	S	92
	S7	Surface antigen	S	93

\*A=in situ hybridisation; S=somatic cell genetics; F=family studies; RE=recombinant DNA techniques; D=deletion mapping.

autosomal recessive disorder in the Caucasian population.<sup>110</sup> The frequency of the mutant gene is estimated to be as high as 5% in some studies.<sup>111</sup> Patients with CF have major symptoms including obstructive pulmonary disease, pancreatic enzyme insufficiency, and raised sweat electrolyte levels. Although a general malfunction in the secretory epithelia seems to be a consistent finding in CF,<sup>112-115a</sup> the basic defect has yet to be discovered. Mapping of the CF locus has been the subject of intensive investigations in the past two years.<sup>51-54 59-63 85 116-125a</sup> Through the mapping of a large number of DNA markers surrounding the disease locus, the most probable chromosome location for CF is within band 7q31.<sup>45 56 62 63 85 86 126</sup> Genetic diagnosis has also been possible with the tightly linked DNA markers.<sup>127-129</sup>

### Chromosome aberrations

There is a large number of documented cases of patients with short arm (7p-) deletions and translocations,<sup>130-147</sup> and most of them have been summarised recently.<sup>147</sup> The majority (60%) of deletions involve the terminal band p22 and about half of these also have band p21 or p15 deleted. Two cases of recurrent spontaneous abortions seem to indicate the involvement of 7p.<sup>148 149</sup> The remaining cases are interstitial deletions of various lengths, spanning between p21 to p13. The most frequent clinical abnormalities associated with 7p deletions are cranial dysmorphism, cleft palate, saddle nose, small, dysplastic, and low set ears, foot malformations, congenital heart disease, and genital malformations. However, the size and location of the chromosome segments involved do not show any obvious correlation with these or other symptoms.

Chromosome abnormalities have also been reported for the long (q) arm of chromosome 7. In contrast to the 7p- condition, the 7q- syndrome exhibits relatively consistent dysmorphic findings.

Patients with terminal 7q deletions<sup>150-162</sup> have growth deficiency, developmental delay, microcephaly, low birth weight, bulbous nasal tip, anomalous auricles, cleft lip/palate, genital malformations in males, and ocular anomalies. One-third of terminal del(7q) patients also have a history of perinatal feeding problems, abnormal EEG with or without seizures, ocular hypertelorism, micrognathia, and chest abnormalities.

Interstitial 7q deletions can be divided into three classes based on the chromosomal regions involved, namely, q11→q21 (or q22), q21→q31 (or q32), and q32→q34, all of which show delayed development as their common feature. The clinical presentations seem to agree well with this classification.<sup>160</sup> In the q11→q21 (or q22) class, patients<sup>139 163-167</sup> generally show feeding problems and some malformation in addition to their developmental delay. The main features in the del(7)(q21→q31 or q32) patients<sup>160 165 168-175</sup> are delayed development, feeding difficulties, ear malformations, simian creases, low birth weight, and an unusual 'cat like' cry. A relatively small number of patients have been studied for the q32→q34 deletion,<sup>160 176 177</sup> but developmental delay, broad nasal bridge, bulbous nasal tip, ocular hypertelorism, and large mouth seem to be consistent within this last class.

Heritable partial trisomy 7q can also be divided on the basis of the chromosomal regions involved,<sup>178</sup> which are q22→q31 or qter,<sup>179-182</sup> q31→qter,<sup>183-186</sup> and q32 (or q33)→qter.<sup>136 178-194</sup> Retardation of development and low set ears are the common clinical features of all three groups but significant differences are noted among them.<sup>178</sup> Strabismus, epicanthus, and frontal bossing seem to be unique to trisomy 7(q22→q31), wide open fontanelle, cleft palate, microretrognathia, and early prenatal death to q31→qter, and kyphoscoliosis and hypotonia to q32 (or q33)→qter. Hypertelorism and small palpebral fissures appear to be common in the first and second groups, and low birth weight, small nose,

and skeletal anomalies in the second and the third groups. Thus, trisomy 7(q31→qter) is more serious than the other two.<sup>178</sup>

Paracentric inversion is infrequent in man but chromosome 7 appears to be affected the most often. This topic has been reviewed recently.<sup>195-196</sup> Most paracentric inversions are apparently harmless and the heterozygous carriers of inv(7) were primarily detected in cases involving infertility, repeated abortions, malformation, and advanced maternal age.<sup>195-207</sup> As in the other chromosome 7 aberrations, the paracentric inversions appear to be non-randomly distributed within this chromosome. The majority of the breakpoints are located in bands q11 and q22, and the inverted segment was q11→q22 in half of the cases.

More recently, uniparental disomy has been proposed as a mechanism for human genetic disease.<sup>208</sup> In the reported case, the presence of two identical copies of the maternal chromosome 7 and the absence of paternal chromosome 7 were thought to be the cause of cystic fibrosis in a patient who also had short stature.

### Somatic cell chromosome aberrations

Monosomy 7 is frequently associated with secondary blood disorders, namely, acute non-lymphocytic leukaemia (ANLL) or myelodysplastic syndrome (MDS), which often occurs in patients undergoing aggressive therapy (irradiation or chemotherapy or both) for a previous malignant disease (for example, Hodgkin's disease).<sup>209-220</sup> A recent review<sup>220</sup> has indicated that more than 90% of these patients show hypodiploidy and monosomy 7 was found in more than 60% of the abnormal cases. There is a highly specific correlation between chromosome 7 abnormality and chemotherapy ( $p < 0.01$ ).<sup>220</sup> A correlation has also been shown between monosomy 7 and haematological disorders in persons exposed to chemical carcinogens in the work place.<sup>221-225</sup> Deletion and translocation of 7q accounts for all of the cases with partial monosomy 7 in the latter category, the common deleted segment being 7q32 or q34.<sup>226</sup> Monosomy 7 and deletion of 7q also seem to be involved in de novo ANLL (or MDS) according to some<sup>227-229</sup> but not all studies.<sup>219-227</sup>

Chromosome 7 abnormalities have been implicated in several other cancer conditions, including ataxia telangiectasia,<sup>230-232</sup> erythroleukaemia,<sup>233-234</sup> gamma heavy chain disease,<sup>235</sup> lung carcinoid tumour,<sup>236</sup> prostate carcinoma,<sup>237</sup> transitional cell carcinoma of the ureter,<sup>238</sup> bilateral renal oncocytoma,<sup>239</sup> and malignant melanoma.<sup>240-243</sup> In vitro cellular studies suggest that chromosome 7 is particularly unstable and vulnerable.<sup>244-246</sup> In-

terestingly, the translocation and deletion breakpoints discussed above seem to coincide well with the fragile sites observed on this chromosome, namely, FRA7A (rare, 7p11.2<sup>247-248</sup>), FRA7B (7p22<sup>249</sup>), FRA7C (7p14.2<sup>249</sup>), FRA7D (7p13<sup>250-251</sup>), FRA7E (7p21.2<sup>249</sup>), FRA7F (7q22<sup>250</sup>), FRA7G (7q31.2<sup>249-251</sup>), and FRA7H (7q32.3<sup>249-250</sup>). In a recent classification of fragile sites, 7q32 is listed as one of the most common fragile sites.<sup>252</sup>

### Oncogenes

Direct involvement of genes on chromosome 7 in tumour formation or malignancy has been suggested. Enhanced expression of the epidermal growth factor (EGF) receptor (cellular homologue of the viral *erbB* gene) in malignant melanoma,<sup>253</sup> epidermal carcinoma,<sup>254-257</sup> pancreatic carcinoma,<sup>258</sup> and glioblastoma<sup>259</sup> cell lines has been correlated with 7p abnormalities and polysomy 7. A strong correlation has been observed between the presence of a 7q abnormality and the expression of the T cell growth factor receptor (Tac) in a group of patients with T cell leukaemia.<sup>260</sup> The possibility of activation of the Tac gene via the T cell receptor  $\beta$  chain gene has been proposed. Chromosome 7 abnormalities have also been detected in some patients with chronic T lymphocyte tumours, but the breaks are located in different bands.<sup>261</sup>

The *met* proto-oncogene, located within the 7q31 region,<sup>56</sup> was shown to have transforming potential in vitro when activated.<sup>55</sup> The transforming activity was first discovered in a mouse NIH/3T3 cell transformation assay with DNA isolated from a MNNG treated human osteosarcoma (HOS) cell line,<sup>262</sup> and it is therefore unlike most other proto-oncogenes, which were initially identified through viral oncogenes. DNA sequence analysis showed that the *met* gene belongs to the tyrosine kinase gene family and is closely related to the human insulin receptor and the viral *abl* gene.<sup>56</sup> Subsequent studies revealed that the activation of *met* in the MNNG-HOS cell line involved a gene fusion event which resulted in the replacement of the 5' portion of the proto-oncogene by sequences originating from chromosome 1.<sup>263-264</sup> The normal cellular function of *met* is as yet unknown.

The integration of the simian virus 40 genome in human chromosome 7 has been associated with cellular transformation.<sup>265</sup> The site of viral integration appears to be non-random<sup>265</sup> and maps to region 7q31.<sup>266</sup> Whether the transformation is the result of viral gene (T antigen) expression or disruption of cellular genes remains to be investigated.



### Gene loci previously mapped to chromosome 7

There are a number of gene loci previously assigned to chromosome 7 but subsequently reassigned to a different chromosome. The two classical markers, the Colton (Co) and the Kidd (Jk) blood groups, were thought to be on chromosome 7 based on several lines of direct and indirect evidence.<sup>151 267 268</sup> Recent data suggest that they are located elsewhere<sup>160</sup> and probably unlinked (J Mohr, 1987, personal communication).

The mapping of the histone genes is interesting. Unlike the sea urchin and *Drosophila* histone genes which are clustered and tandemly repeated,<sup>269</sup> the human genes,<sup>270 271</sup> as well as those in yeast,<sup>272</sup> mouse,<sup>273 274</sup> and chicken,<sup>275 276</sup> are clustered but have no apparent repeats. Previous investigations, based on in situ hybridisation with probes derived from a histone mRNA enriched RNA fraction from human cells or sea urchin histone cDNA, showed that the human histone genes are either located within band 7q22 or 7q32→36.<sup>277 278</sup> However, when specific gene probes were used, these genes were found to be in several different chromosome regions; one cluster containing H3 and H4 has been localised to 1cen→q25<sup>279</sup> and other clusters containing core (H3, H4, H2A, and H2B) alone or core together with H1 have been assigned to 1cen→q31, 6p12→q16, and 12p11.2→q21.<sup>11</sup> A small number of grains was observed between the 7pter→q11 when a H1 specific probe was used in the latter in situ hybridisation experiment, but no signal was detectable by gel blot hybridisation analysis with two somatic hybrid cell lines, each containing chromosome 7 as the only human chromosome. The sequences detected on chromosome 7 by in situ hybridisation may represent cross hybridisation of other histone genes, a possibility that remains to be tested.

### Genetic linkage map

Since the localisation of CF to 7q31, joint studies have been carried out to examine the linkage relationship among markers in this region.<sup>117 280</sup> Six loci have been analysed further and the gene order with greatest statistical support is *COLIA2-D7S13-D7S16-met-D7S8-TCRB*.<sup>280</sup> The most likely location of the CF gene on this map is between *met* and *D7S8*. A linkage map has also been generated for almost the entire length of chromosome 7 with randomly isolated DNA markers<sup>281</sup>; the map spans 250 cM in females and 170 cM in males, values much greater than previously estimated.<sup>2</sup> The most significant differences between the male and female genetic map appears to be within the portion of 7q bound by *COLIA2* and *TCRB*.<sup>280 281</sup>

### Comparative mapping

The use of conserved linkage groups among mammals and primates has been an alternative approach to understanding the human gene map and evolution of chromosomes. Comparative mapping studies showed that human chromosome 7 markers are distributed over six different mouse chromosomes and that the mouse chromosome 5 and 6 sequences are found in both the long and short arms of human chromosome 7<sup>282 283</sup> (table) (S Naylor, 1987, personal communication).

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## Genetic markers on chromosome 7.

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