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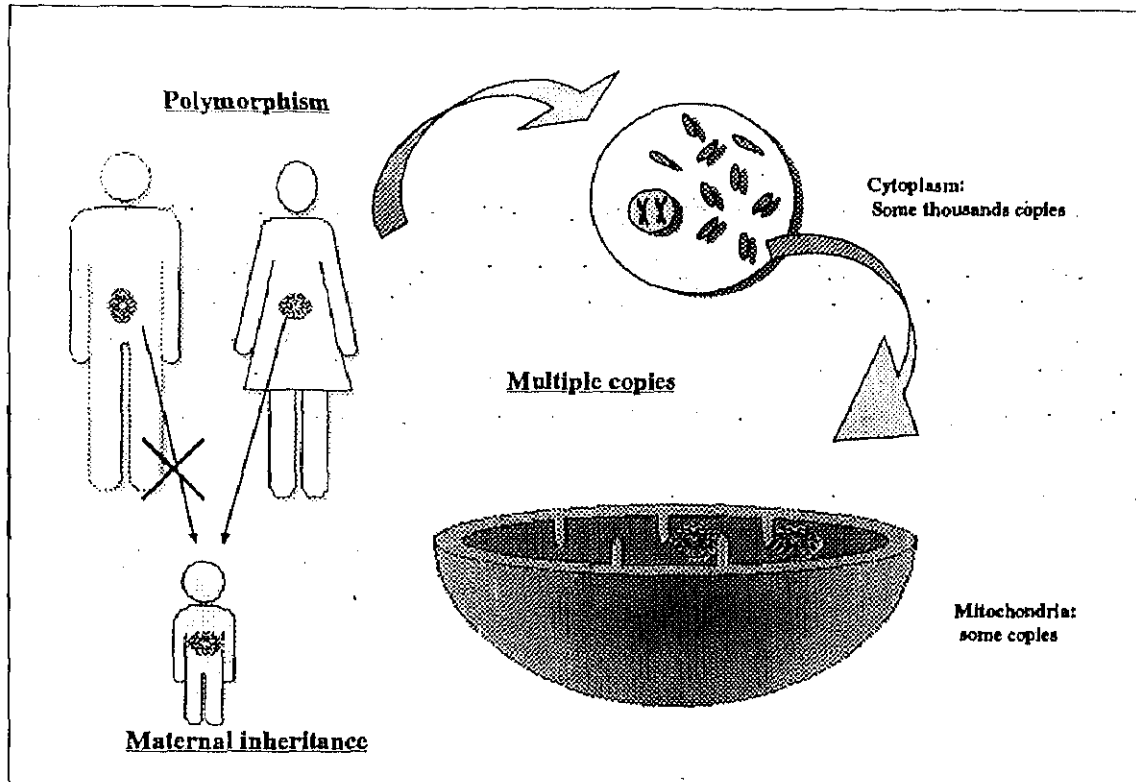


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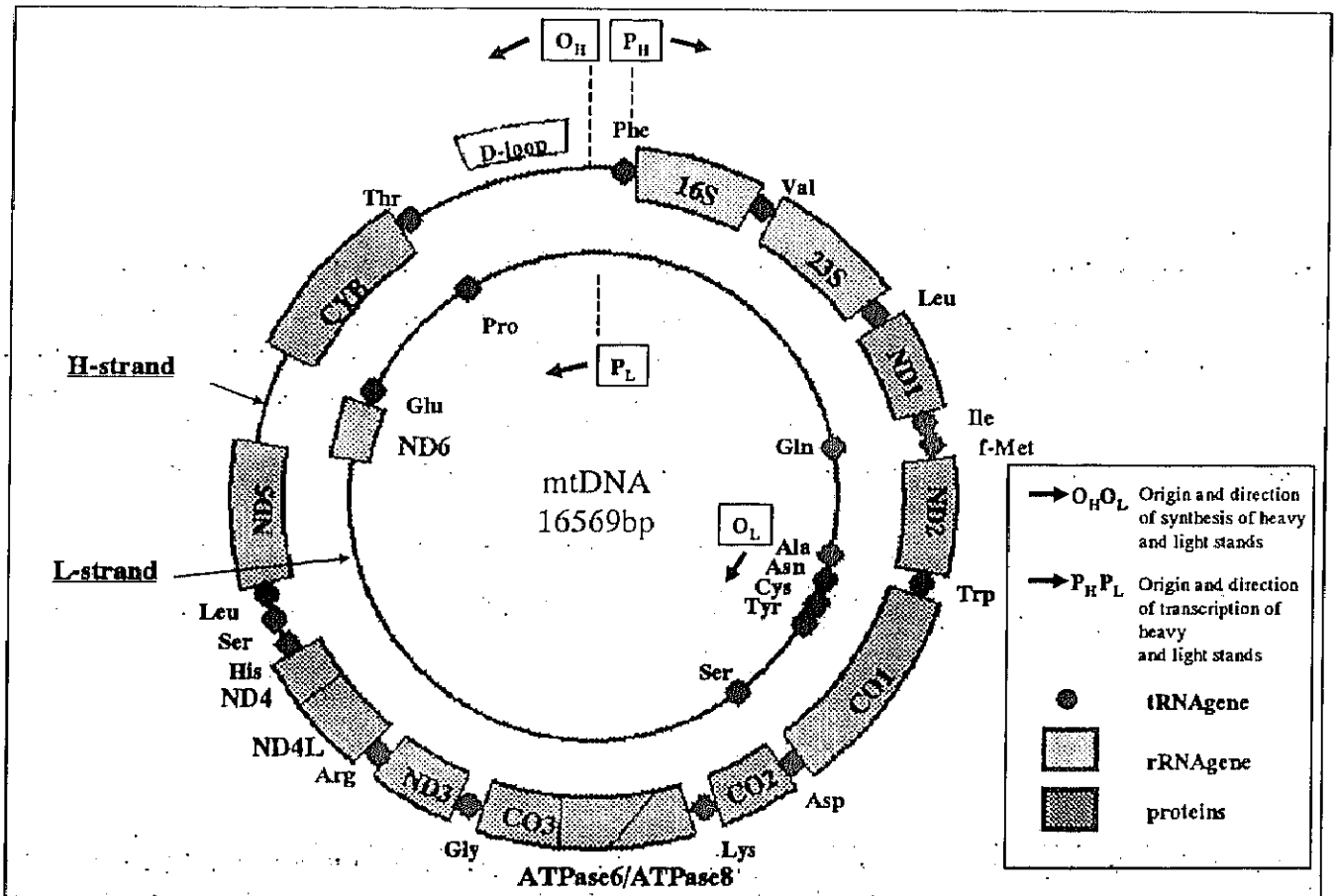
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**Figure 1 Feature of mtDNA**



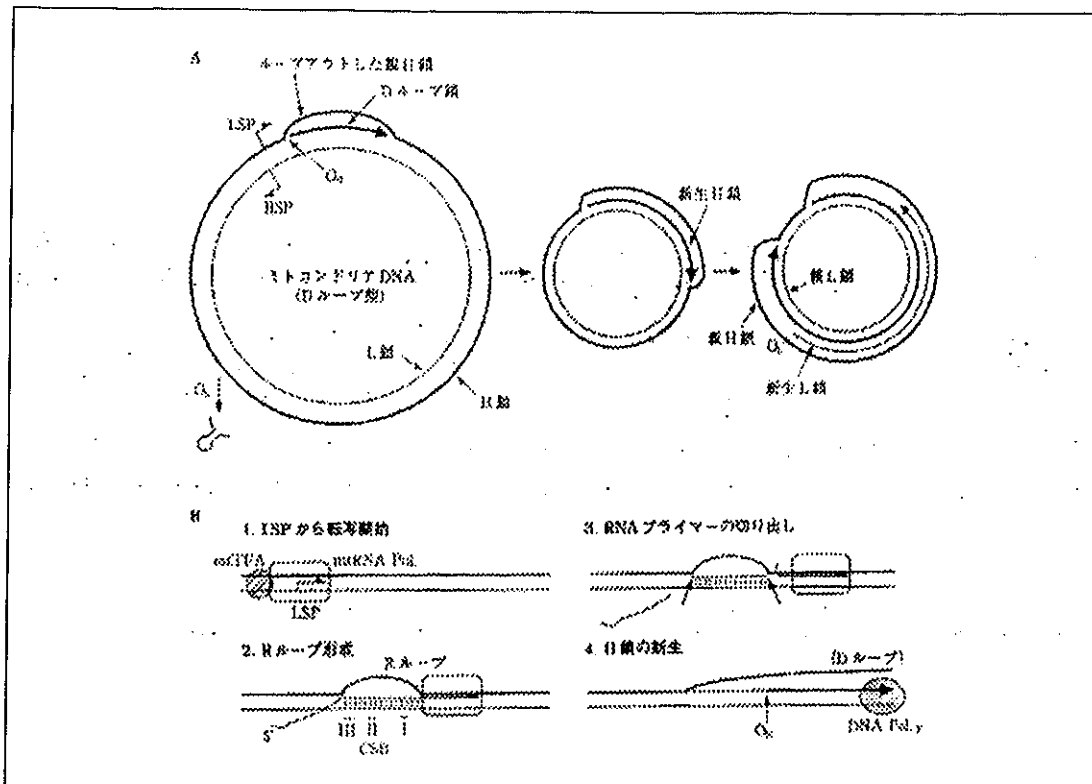
**Figure 2: mtDNA map**



**Table 1: human mtDNA codon** (tRNA anti-codon)

Phe (GAA)	UUU UUC		Thr (UGU)	ACU ACC ACA ACG		Asp (GUC)	GAU GAC
Leu (1) (UAA)	UUA UUG		Ala (UGU)	GCU GCC GCA GCG		Glu (UUC)	GAA GAG
Leu (2) (UAG)	CUU CUC CUA CUG		Tyr (GUA)	UAU UAC		Cys (GCA)	UGU UGC
Ile (GAU)	AUU AUC		Ter	UAA UAG		Trp (UCA)	UGA UGG
Met (CAU)	AUA AUG		His (GUG)	CAU CAC		Arg (UCG)	CGU CGC CGA CGG
Val (UAC)	GUU GUC GUA GUG		Gln (UUG)	CAA CAG		Ser (2) (GCU)	AGU AGC
Ser (1) (UCN)	UCU UCC UCA UCG		Asn (GUU)	AAU AAC		Ter	AGA AGG
Pro (UGG)	CCU CCC CCA CCG		Lys (UUU)	AAA AAG		Gly (UCC)	GGU GGC GGA GGG

**Figure 3A: D-ループ鎖をもつmtDNAとstrand-asynchronous複製途中のmtDNA**  
**3B: H鎖新生開始のモデル(文献44より一部改編)**



A; H鎖、L鎖それぞれの転写開始点はHSP、LSPで表す。H鎖、L鎖それぞれの複製開始点は $O_H$ 、 $O_L$ で表す。 $O_H$ から複製された約700bpのDNAフラグメントである、Dループ鎖から親L鎖を鋳型としたH鎖新生が進み、 $O_L$ の部分の親H鎖が一本鎖になると $O_L$ はステムループ構造をとり、複製開始のシグナルとなると考えられる。

B: L鎖の転写開始とH鎖新生開始が一連におこなわれている。

(1)LSPからRNA新生が起こる。転写因子mtTFAとmtRNA Pol.が主にかかわっている。

(2)RNA新生が進むとCSBsの存在する領域と、RNAが安定なRNA-DNAハイブリッドが形成され、親H鎖がほどかれたままのRループ構造が形成される。RNA-DNAハイブリッドは単純な二本鎖らせん構造をとっているわけではなく、RNAの一部の領域が一本鎖で特殊な構造をとっていることが示唆されている。

(3)そしてこのRNA-DNAハイブリッド構造がRNaseMRPによって認識されて適切な位置でRNAが切断され、DNA伸長のためのRNAプライマーとなる。(4)RNAプライマーを用いて、DNA Pol.  $\gamma$ がDNA鎖を伸長していく。DNA複製開始点はCSB Iのすぐ下流である。

**Figure 4: Control region of mtDNA**

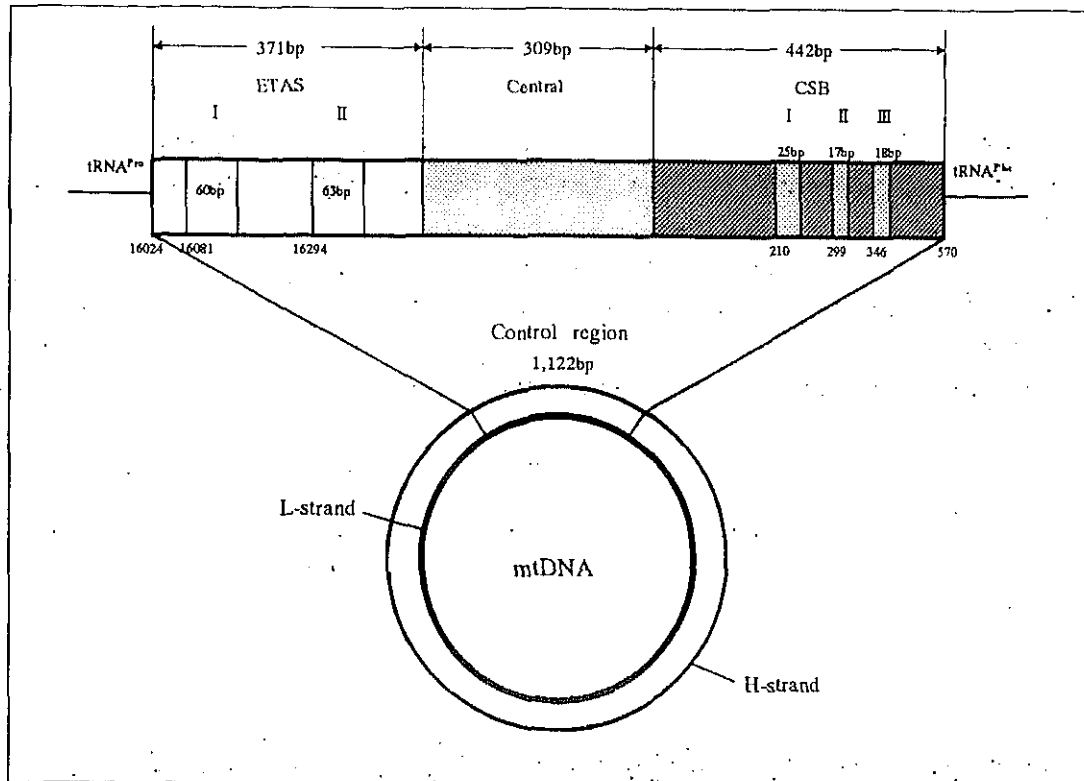
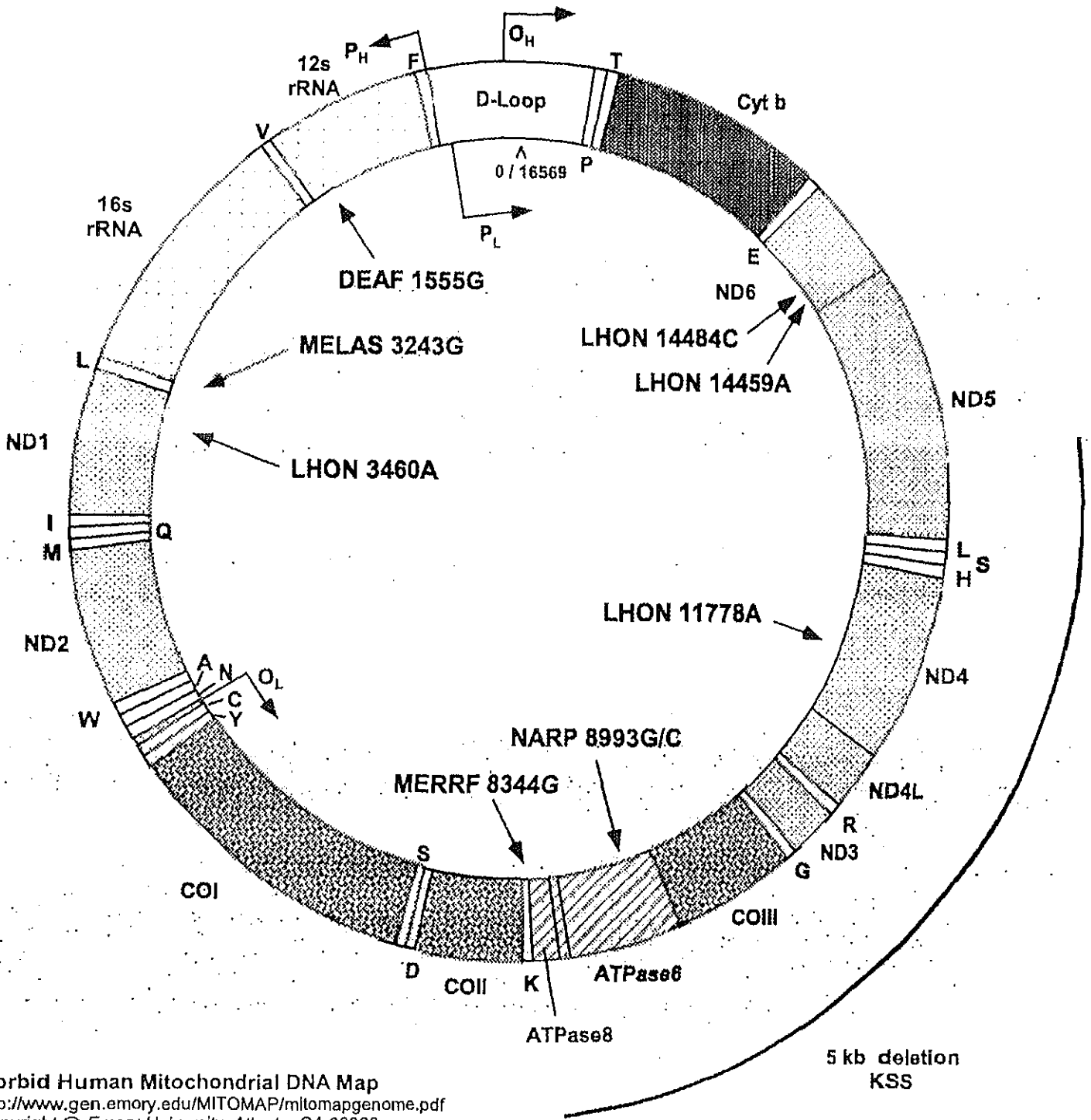


Figure 5: Human mtDNA map and mitochondrial disease point (MITOMAP<sup>75</sup>)



Morbid Human Mitochondrial DNA Map  
<http://www.gen.emory.edu/MITOMAP/mitomapgenome.pdf>  
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Table 4: Location and rate of main SNPs in HV1 and HV2 of mtDNA

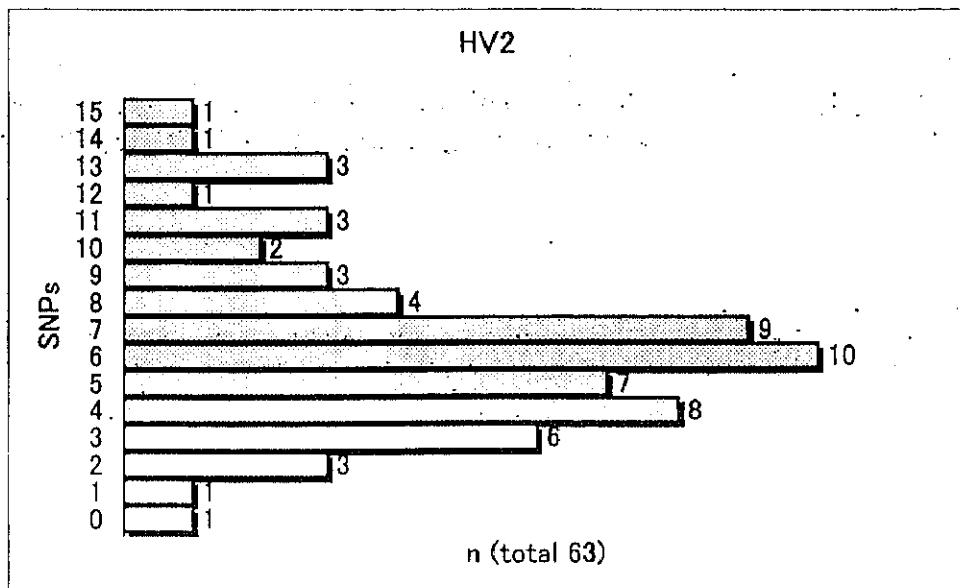
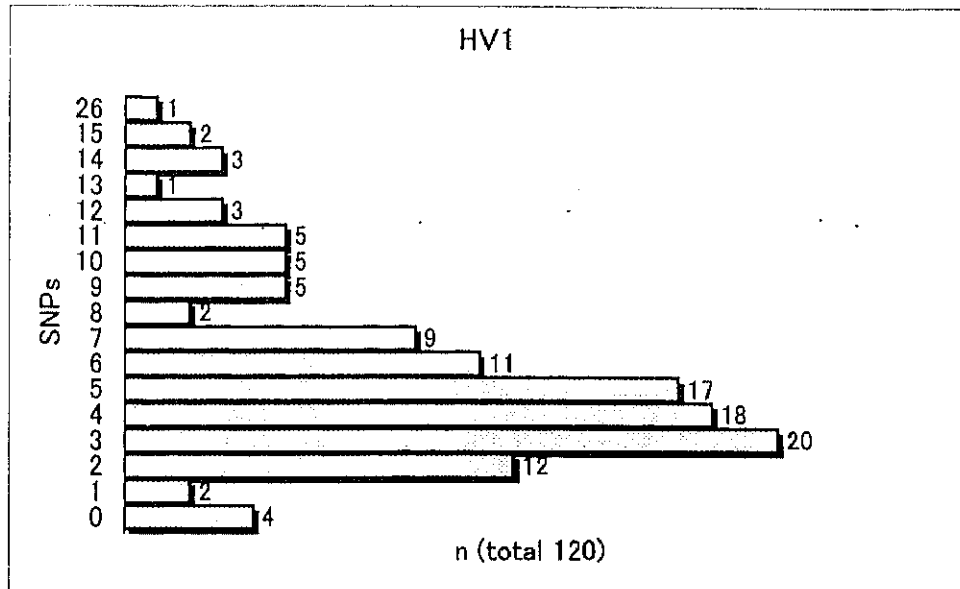
HV1

nps		n	rate(%)	reference
16223	C → T	52	43.33	
16183	A → C	32	26.67	
16129	G → A	27	22.50	
16189	T → C	26	21.67	
16182	A → C	24	20.00	
16127	A → G	19	15.83	
16362	T → C	16	13.33	
16111	C → T	16	13.33	
16091	A → T	16	13.33	-
16362	T → G	14	11.67	-
16172	T → C	12	10.00	
16320	C → G	12	10.00	-
16325	T → G	11	9.17	-
16214	C → G	10	8.33	-

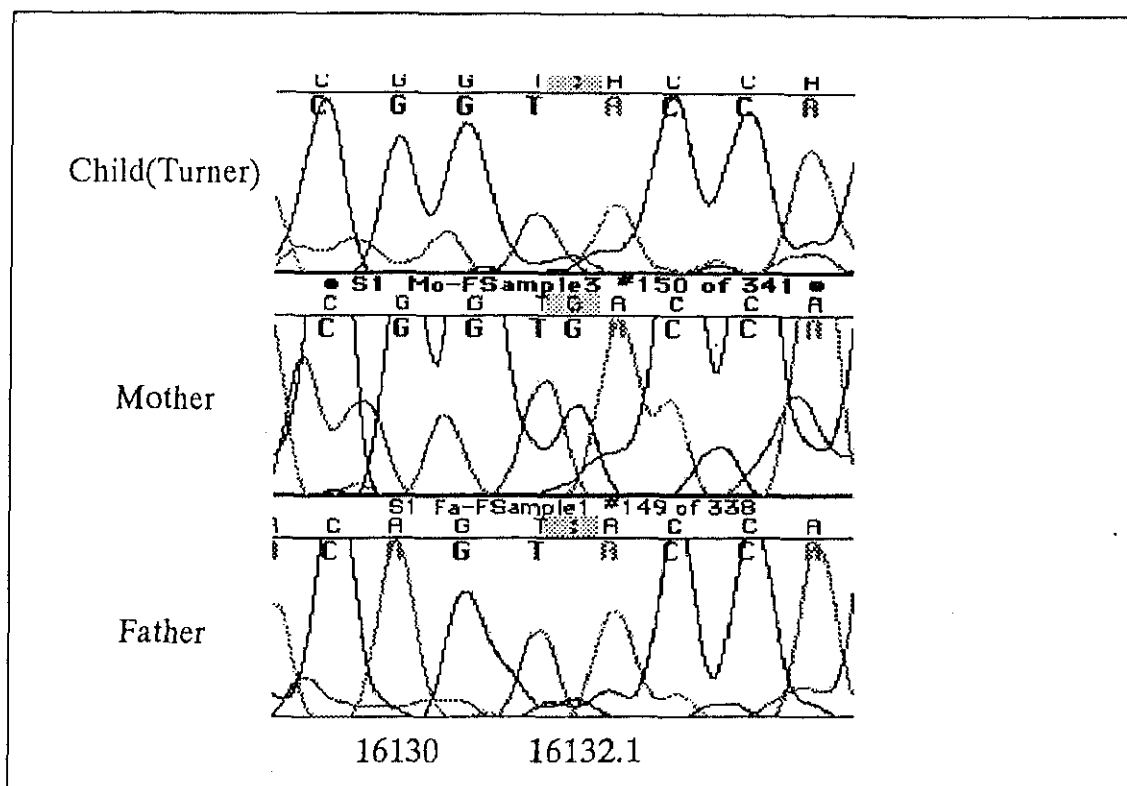
HV2

nps		n	rate(%)	reference
73	A → G	55	87.30	
263	A → G	55	87.30	
330	C → G	28	44.44	-
324	C → G	25	39.68	-
381	C → G	17	26.98	-
348	C → G	13	20.63	-
377	C → A	13	20.63	-

Figure 6: Number of SNPs in HV1 and HV2 of mtDNA each of 120 and 63 individuals.



**Figure 7: Sequencing of mtDNA at nps 16129-16136 in Turner syndrome and her parents**



**Table 5: Location and number of all SNPs ,specially no report regions in HV1 of mtDNA.**

nps	16091	92	93	95	96	97	98	102	104	105	106	107	110	111	120	122	123	124	126	127	128	129
Anderson	A	T	T	C	G	T	A	T	C	T	G	C	G	C	A	A	T	T	T	A	C	G
mutation&number	T16	C2	C3	T1	C1	C2	T7	C1	T1	C3	A2 C1	T3	A7	T16	T7	T2 C1 G1	C2	C1	C3	G19	G2	A27 C1
nps	131	136	137	138	139	140	144	145	148	155	156	158	162	163	164	165	169	170	171	172	173	178
Anderson	T	T	A	A	A	T	T	G	C	A	G	A	A	A	A	A	C	A	A	T	C	T
SNPs and number	C1	G1	G6	T1	T3	C8	C1 G2	A1	T1	G3	A1 C2	G1	G2	G1	G3	T1	T1	G8	G1	C12	T1	C1
nps	182	183	185	186	189	192	194	196	197	199	200	201	203	204	205	207	208	209	210	211	214	216
Anderson	A	A	C	C	T	C	A	G	C	T	A	C	A	G	C	A	G	T	A	C	C	A
SNPs and number	C24	C32	T1	T3	C26	T4	C4 T1	T1	G1	A1	T1 C1	A3	G2	A1	A1	G1	T2	C7	G2	A1	T1	G2 A1 T2
nps	217	218	220	221	223	227	228	230	231	234	241	242	243	244	245	247	248	250	254	255	256	257
Anderson	T	C	A	C	C	A	C	A	T	C	A	C	T	G	C	A	C	C	A	G	C	C
SNPs and number	C1	T1	G2	G1	T52	G3 T1	T1	G1	C2	G1	G2	T1	C2	C1 A2	T5 G3 A2	G1	T1	G1	G1	A1	G4	A1
nps	258	260	263	264	265	266	269	270	271	272	275	277	278	285	286	287	288	289	290	291	292	295
Anderson	A	C	T	C	A	C	A	C	T	A	A	A	C	A	C	C	T	A	C	C	C	C
SNPs and number	C1	T1	C1	T2	C2	A3 T4	C1	A4	C2	G1	G3 T1	C1 G1	T8	C3 G1	T1 G1	T1	C4	C1 T1	T4	T3	A1	A1 T2
nps	296	297	298	299	301	304	305	309	311	312	313	316	317	318	319	320	324	325	326	327	329	330
Anderson	C	T	T	A	C	T	A	A	T	A	C	A	A	A	G	C	T	T	A	C	G	T
SNPs and number	G1	C2	C3 A2	G1	T2 G1	G2 C7	T1	G2	C7 G2	T2	T5	G1 T5	G1	G2	A6	G12 A1	C7 G1	C1 G1	T1 C1	A1 T3	C2	G2
nps	331	335	337	343	344	345	347	351	354	355	356	357	358	359	360	362	371	375	376	380		
Anderson	A	A	C	A	C	A	T	A	C	C	T	T	C	T	C	T	A	C	C	C		
SNPs and number	G6	G3 T3	G6	G1	G1 A1	G1 C1	G4	T1	A1	A4	A1	C2	T4	C2	T3 G1	C16 G14	T2 G3	A1	A1	T1		

no report SNPs

**Table 6: Location and number of all SNPs ,specially no report regions in HV2 of mtDNA.**

nps	33	41	48	56	64	65	73	75	79	80	86	89	90	91	98	108	133	134	135	140	141	143	144	146	148	150
Anderson	C	C	C	A	C	T	A	G	G	C	C	T	G	C	C	A	T	T	T	C	C	G	C	T	A	C
SNPs and number	T1	A1	A2	T2	T1	G1	G55	A1	A1	G2	A3	C1	C1	T5	T1	T1	C1	G2	G1	G1	G1	A2	A2	C3	T1	T7
			G1				C1			T1	G2	A1		A2				G1								
nps	151	152	173	176	186	194	195	199	202	204	211	219	222	227	228	234	235	241	249	261	263	275	276	277	280	281
Anderson	C	T	T	A	C	C	T	T	A	T	A	A	C	A	G	A	A	A	A	C	A	G	A	C	C	A
SNPs and number	T1	C8	C1	C1	G2	T4	C2	C1	T1	C1	C1	C1	T1	G1	A1	G1	G6	T1	C1	G5	G55	A1	C1	G1	A1	T1
		G1												C5				C1		T2	T3		G1			
nps	283	284	287	288	290	291	301	310	316	317	319	322	323	324	327	330	332	335	336	337	338	340	343	345	348	359
Anderson	A	A	A	A	A	A	A	T	G	C	T	G	G	C	C	C	C	A	A	A	C	C	C	C	C	A
SNPs and number	T1	G1	T3	C1	T3	T3	C1	C6	C2	G7	C1	A1	A1	G25	A1	G28	T1	T1	T1	T1	A1	A1	T1	T1	G13	T1
		T2								T13				A1		T1	A2									T2
nps	360	363	365	367	369	373	374	375	377	379	381	384	386	390	393											
Anderson	A	A	A	A	C	A	A	C	C	A	C	A	C	A	T											
SNPs and number	T1	C1	T2	G1	A1	T1	G1	A1	A13	C1	G17	T1	T2	G1	C1											
											A1					T1										

no report SNPs



Table 7: mtDNA-HV haplotypes in Klinefelter syndrome and his mother

**HV1**

	nps	16042	16060	16089	16208	16384
Anderson		G	G	G	G	G
Child (Klinefelter)		A	G/T	G/T	T	A
Mother		A	T	T	G	C

**HV2**

	nps	80	120	126	223	227	254	263	299
Anderson		C	C	A	T	A	T	A	C
Child (Klinefelter)		G	C	C	T	A	T	G	C
Mother		T	G	A	G	G	G	G	G

Table 8: mtDNA-HV haplotypes in Turner syndrome and her parents

**HV1**

	nps	16129	16136	16183	16095-6	16110-11	16129-30	16150-1
Anderson		G	T	A	CG	GC	GG	CC
Father		A	T	A	CG	GC	GG	CC
Child (Turner )		G	C	C	CG	GC	GG	CC
Mother		G	C	C	CGG	GAC	GGG	CCG

**HV2**

	nps	73	199	202	207	263
Anderson		A	T	A	G	A
Father		G	T	G	G	G
Child (Turner )		G	C	G	A	G
Mother		G	C	G	A	G

**Table 9: Frequencies of common polymorphisms in control region in different human population groups.**

	16223	16362
	C→T	T→C
Aboriginal Siberians <sup>92</sup>	93.8%	50.0%
Aboriginal Taiwanese <sup>93</sup>	72.9%	46.4%
African <sup>94</sup>	87.4%	15.8%
Amerindian-Blazil <sup>95</sup>	73.0%	61.0%
Amerindian-Costa Rica <sup>96</sup>	96.3%	92.6%
Caucasian-Bulgaria <sup>97</sup>	6.5%	3.2%
Caucasian-France <sup>98</sup>	10.0%	6.0%
Caucasian-Germany <sup>99</sup>	5.0%	4.0%
Caucasian-Swiss Alps <sup>100</sup>	0.0%	12.9%
Caucasian-UK <sup>101</sup>	8.0%	5.0%
Chinese-Hong Kong <sup>102</sup>	55.0%	20.0%
Indian <sup>103</sup>	64.6%	19.4%
Japanese <sup>76</sup>	78.0%	45.0%
Kazakhs-Central Asia <sup>104</sup>	50.9%	32.7%
Korean <sup>105</sup>	78.0%	39.9%
Mapuche <sup>106</sup>	61.5%	38.5%
Mongolian <sup>107</sup>	56.5%	34.3%
Nuu-Chah-Nulth <sup>108</sup>	82.1%	64.2%
Turks <sup>109</sup>	17.3%	3.5%
Saami-Norway <sup>97</sup>	4.8%	3.2%
Sary-Tash K-Central Asia <sup>105</sup>	48.9%	29.8%
this study	43.3%	11.7%

Table 9: (Continued)

	73	263
	A→G	A→G
Aboriginal Taiwanese <sup>98</sup>	100.0%	100.0%
African <sup>110</sup>	100.0%	NA
Amerindian-Brazil <sup>111</sup>	94.0%	72.0%
Amerindian-Costa Rica <sup>96</sup>	62.9%	100.0%
Caucasian-Bulgaria <sup>110</sup>	67.8%	100.0%
Caucasian-France <sup>98</sup>	50.0%	100.0%
Caucasian-Italy <sup>112</sup>	56.3%	100.0%
Caucasian-UK <sup>101</sup>	44.0%	99.0%
Indian <sup>103</sup>	98.0%	100.0%
Japanese <sup>76</sup>	100.0%	99.0%
Korean <sup>106</sup>	99.1%	98.0%
Saami-Norway <sup>111</sup>	59.7%	100.0%
Turks <sup>97</sup>	62.0%	100.0%
this study	87.3%	87.3%

NA: not available