Mitochondrial DNA depletion, mitochondrial mutations and high TFAM expression in hepatocellular carcinoma

SUPPLEMENTARY MATERIALS

Supplementary Table 1: Information of 86 HCC subjects and TFAM expression

See Supplementary File 1

Supplementary Table 2: Summary information of substitutions in 637 mtDNA sites among 86 HCC pairs

See Supplementary File 2

Supplementary Table 3: Overview of mtDNA variants in 86 HCC pairs

Region/gene	Var no.(%)	Transitions	Transversions	del	ins	Non-syn	Syn	novel Var
Control region	142 (22.29%)	112	16	6	8			6
other non-coding	9 (1.41%)	5	0	2	2			0
12S rRNA	23 (3.61%)	21	1	0	1			1
16S rRNA	25 (3.92%)	24	1	0	0			2
tRNAs	35 (5.49%)	32	2	0	1			5
MT-ND1	39 (6.12%)	37	2	0	0	14	25	2
MT-ND2	31 (4.87%)	28	3	0	0	13	18	1
MT-ND3	12 (1.88%)	12	0	0	0	5	7	0
MT-ND4	42 (6.59%)	40	2	0	0	10	32	1
MT-ND4L	7 (1.10%)	6	1	0	0	4	3	0
MT-ND5	71 (11.15%)	66	4	1	0	30	41	5
MT-ND6	13 (2.04%)	13	0	0	0	3	10	0
MT-CO1	40 (6.28%)	38	2	0	0	10	30	2
MT-CO2	24 (3.77%)	24	0	0	0	4	20	0
MT-CO3	29 (4.55%)	28	1	0	0	8	21	0
MT-CYB	59 (9.26%)	53	5	0	1	24	35	2
MT-ATP6	26 (4.08%)	25	0	0	1	12	14	1
MT-ATP8	10 (1.57%)	8	2	0	0	5	5	0
Total	637	572	42	9	14	142	261	28

Var: variants; Syn: synonymous.

Supplementary Table 4: Novel variants in HCC pairs

Region/ Gene	Position	Replacement	Codon	Amino change	17 species conservation	haplotype	T/N	Hom/Het
D-loop	157	T-C				A4	T	Hom
	294-341	48bp-del				F1a1c	T	Hom
	394	C-A				F1a1c	T	Hom
	528	T-C				F1a1d	T	Hom
	540	A-C				F1a1d	T	Hom
	568	C-CC				G2a2	TN	Hom
MT-TF	617	G-A				F1a1c	T	Hom
12S rRNA	955	A-AC				M7c3b	N	Hom
16S rRNA	2864	T-C	1194			F1a1c	TN	Hom
	3222	C-A	1551			M7c3	TN	Hom
MT-ND1	3877	T-C	191	Ala-Pro	17/17	F1a1c	T	Hom
	4153	G-A	283	Asp-Asn	17/17	A4	TN	Hom
MT-TM	4468	T-A	72			M7b1	TN	Hom
MT-ND2	4963	G-A	165	Gly-Asp	17/17	F1a1c	T	Hom
MT-CO1	5942	A-G	13	Syn	17/17	F1a	TN	Hom
	6582	G-A	227	Asp-Asn	17/17	F1a1c	T	Hom
MT-TS1	7509	T-TT	6			D5a2a	TN	Hom
MT-ATP6	8628	C-CC	34	Stop-gain	12/17	D4i	T	Hom
MT-TG	10041	A-C	55			F1a1d/A17	TN	Hom
MT-ND4	11226	G-A	156	Gly-Asp	17/17	F1a1c	T	Hom
MT-TS2	12214	C-T				M7	TN	Hom
MT-ND5	13267	G-T	311	Gly-Trp	17/17	F1a1c	T	Hom
	13475	T-Del	380	Leu-Stop	17/17	A4/M7c3	T/T	Hom
	13603	A-G	423	Ser-Gly	17/17	F1a1a/F1a1c/N9a	T/TN/T	Hom
	13718	G-A	461	Ser-Asn	17/17	F1a1	T	Hom
	13754	C-A	473	Ser-Tyr	17/17	F8a2	TN	Hom
MT-CYB	14765	A-T	7	Thr-Ser	8/17	A4/N9a1/D4i	N/N/T	Hom
	14984	C-CA	80	Stop-gain	15/17	F2a	T	Hom

T/N: T for tumor tissue, N for non-tumor tissue.
Hom/Het:Hom for homoplasmy, Het for heteroplasmy.

Supplementary Table 5: Putative pathogenic variants identified in mtDNA protein coding region in HCC samples

See Supplementary File 3

Supplementary Table 6: Heteroplasmic variants in HCC pairs

Gene	Position	Replacement	aa change	Tumor tissue		Non-tumor tissue	
				Het (No.)	Hom (No.)	Het (No.)	Hom (No.)
D-loop	16213	G-A		A/G (1)	*A (3)	0	A (2)
D-loop	16261	C-T		0	T (6)	C/T	T (6)
	16311	T-C		0	C (5)	T/C	C (5)
MT-RNR2	1736	A-G		0	G (8)	A/G	G (6)
MT-ND2	4883	C-T	Thr-Ala	0	T (16)	C/T	T (16)
MT-ND5	12092	C-A	Leu-Ile	0	0	C/A	0
MT-ND5	13676	A-G	Asn - Ser	A/G (1)	0	0	0

*A (3): adenine was occurred in 3 tumor tissues.