

Table 1: Patient characteristics

Patient #	Normal range	P1	P2	P3	P4	P5	P6	P7	P8
Age (y)		21	16	18	19	21	21	15	11
Sex (F/M)		M	F	M	F	F	F	M	F
Pathology		FHBL-SD1	FHBL-SD1	FHBL-SD1	FHBL-SD3	FHBL-SD3	FHBL-SD3	FHBL-SD3	FHBL-SD3
Mutation variant		NM_000253.4 (MTTP):c.582 C>A	NM_000253.4 (MTTP):c.582 C>A	NM_000253.3 (MTTP):c.59_61+14del	c.1-4482_58+1406 del5946 ins15pb	c.1-4482_58+1406 del5946 ins15pb	c.364 G>T	c.364 G>T	c.554 G>T
Amino acid change		p.(Cys194*)	p.(Cys194*)	p.(Lys20Thrfs*3)	p.(Met1_His43 del)	p.(Met1_His43 del)	p.(Glu122X)	p.(Glu122X)	p.(Gly185Val)
Status		homozygous	homozygous	homozygous	homozygous	homozygous	homozygous	homozygous	homozygous
Weight (kg)		49	45	53	64.6	52.6	60	39	30
Height (cm)		161	155	155	161.6	170.6	167	153	134
BMI (kg/m ²)		18.9	18.7	22.1	24.7	18.1	21.5	16.7	16.7
Treatment									
Vitamin A		50.000UI 2 pills /d	50.000UI 2 pills /d	50.000UI 3 pills / w	50.000UI 1 pill / 15d	50.000UI 1 pill / w	50.000UI 3 pills x 2/d	50.000UI 3 pills x 2/d	50.000UI 1 pill / day
Vitamin E		α -tocopherol acetate 500 mg 3 pills x 2 /d	α -tocopherol acetate 500 mg 3 pills x 2 /d	α -tocopherol acetate 500 mg 3 pills x 2 /d	α -tocopherol acetate 500 mg 3 pills x 2 /d	α -tocopherol acetate 500 mg 3 pills x 2 /d	α -tocopherol acetate 500 mg 3 pills x 2 /d	α -tocopherol acetate 500 mg 3 pills x 2 /d	α -tocopherol acetate 500 mg 3 pills x 2 /d
Vitamin D ₃		100 000 UI 1 /m	100 000 UI 1 /m	100 000 UI 1 /m	100 000 UI 1 /m	100 000 UI 1 /m	100 000 UI 1 /m	100 000 UI 1 /m	100 000 UI 1 /m
Vitamin K ₁		10 mg 2 /w	10 mg 2 /w	10 mg 2 /w	10 mg 1 / w	10 mg 1 / w	10 mg 2 / w	10 mg 1/ w	10 mg 1 / w
Omega-3 fatty acids		1680 mg/d	1680 mg/d	1680 mg/d	2250 mg	2250 mg	1680 mg	1680 mg	1680 mg
Biochemical parameters									
Total cholesterol (mmol/L)	3.5-7	1.07	1.04	1.03	3.43	2.48	1.57	1.76	2
LDL cholesterol (mmol/L)	<4.1	0.03	NM	NM	2.45	1.66	0.88	1.03	1.26
HDL cholesterol (mmol/L)	> 1	1.01	1.02	1.03	0.5	0.57	0.49	0.44	0.46
TG (mmol/L)	0.4-1.7	0.06	0.05	0.06	1.06	0.56	0.45	0.65	0.62
ApoB (g/l)	0.55-1.15	<0.03	<0.03	<0.03	0.76	0.41	0.29	0.32	0.4
Vitamin A (μ mol/L)	1.4-3.2	2.66	1.63	1.98	2	2.29	1.16	1.28	1.42
25OH Vitamin D	>50	230	204	79	191	195	19	145	80

(nmol/L)									
Vitamin E (µmol/L)	20-35	1.8	2.6	1	9.5	7.8	2.79	3.83	15.23
ALAT (U/L)	<55	86	46	111	44	54	54	45	35
ASAT (U/L)	5-34	48	52	89	39	58	50	53	42
GGT (U/L)	9-36	93	101	29	13	12	22	48	11
Total bilirubinemia (µmol/L)	3-21	20	15	23	7	14	8	6	<5
CK (U/L)	< 170	56	49	91	536	716	510	487	219
PAL (U/L)	40-150	86	82	72	104	101	85	405	208
Fer (µmol/L)	9-30	30	8	17	10	13	4	12	12
Ferritin (µg/L)	5-204	47	11	34	7	15	5	15	6
Hemoglobin (g/L)	120-160	168	99	115	130	142	98	142	126
Acanthocytes	< 5%	+++	+++	0	0	0	+	+++	0
Prothrombin ratio (%)	> 70%	82	77	97	98	87	59	74	77
Ophthalmic function*		Nal	Nal	Nal	Nal	Nal	Nal	Nal	Nal
Visual evoked potential		Nal	Nal	Nal	Nal	Nal	Nal	Nal	Nal
Electro-retinogram		Nal	Nal	Nal	Nal	Nal	Nal	Nal	Nal
Carotenoids									
α-carotene (nmol/L)		0.00	1.92	4.92	10.87	34.63	3.99	5.75	25.45
(nmol/ mmol chol)		0.00	1.85	4.78	3.17	13.97	2.54	3.27	12.73
β-carotene (nmol/L)		0.00	5.15	5.12	9.57	34.52	5.23	14.50	147.06
(nmol/ mmol chol)		0.00	4.95	4.97	2.79	13.92	3.33	8.24	73.53
Lutein (nmol/L)		3.30	9.95	5.85	6.06	27.70	3.81	5.87	3.75
(nmol/ mmol chol)		3.08	9.57	5.68	1.77	11.17	2.42	3.34	1.87
Zeaxanthin (nmol/L)		0.00	16.80	0.80	2.41	16.64	0.41	3.09	0.73
(nmol/ mmol chol)		0.00	16.16	0.78	0.70	6.71	0.26	1.76	0.37
β-cryptoxanthin (nmol/L)		0.00	36.90	17.92	15.39	36.52	5.74	2.22	10.67
(nmol/ mmol chol)		0.00	35.49	17.40	4.49	14.73	3.66	1.26	5.34
Lycopene (nmol/L)		8.59	2.34	6.82	40.01	76.54	7.73	46.58	37.62
(nmol/ mmol chol)		8.03	2.25	6.62	11.66	30.86	4.92	26.47	18.81