ABCB4 variant is associated with hepatobiliary MR abnormalities in people with low-phospholipid-associated cholelithiasis syndrome

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Table of contents

Table S1: Number of MRCP in LPAC patients

Number of MRCP	No Variant (n=65)	Variant (n=60)	Total
1	42 (42)	27 (27)	69 (69)
2	8 (16)	10 (20)	18 (36)
3	3 (9)	5 (15)	8 (24)
4	4 (16)	8 (32)	12 (48)
5	3 (15)	0 (0)	3 (15)
More than 5	5 (43)	10 (77)	15 (120)

Table S2: Radiological follow up according to ABCB4 gene status

	ABCB4 variant (n= 31)	No ABCB4 variant (n= 16)	P
Improvement (14)	11 (35 %)	3 (18 %)	
Stability (21)	14 (45 %)	7 (43 %)	NS
Worsening (12)	6 (19 %)	6 (36 %)	

Table S3 - Genetic status in the 60 patients with ABCB4 variant

CASE	SEX	AGE	LOCATION AND NUCLEOTIDE CHANGES	SEQUENCE VARATIONS	IMPORTANCE OF SEQUENCE VARIATION *	STATUS
1	F	43	c.2800G>A	p.Ala934Thr	1	HTZ
2	F	24	c.140G>A & c.1217G>A	p.Arg47Gln & p.Arg406Gln	1	HTZ & HTZ
3	F	39	c.2800G>A	p.Ala934Thr	1	HTZ
4	M	51	c.857C>T	p.Ala286Val	2	HTZ
5	F	53	c.1769G>A	p.Arg590Gln	1	HTZ
6	F	64	c.2363G>A	p.Arg788Gln	2	HTZ
7	F	21	c.139C>T & c.823A>G	p.Arg47* & p.Thr175Ala	1	HTZ & HTZ
8	F	33	c.2869C>T	p.Arg957*	1	HTZ
9	F	34	c.3258C>A	p.Tyr1086*	1	HTZ
10	F	65	c.2471T>G	p.Val824Gly	2	HTZ
11	F	41	c;2363G>A	p.Arg788Gln	2	HTZ
12	M	76	c.1769G>A & c.2800G>A	p.Arg590Gln & p.Ala934Thr	1	HTZ & HTZ
13	F	28	c.1015dupT	p.Asp355*	1	HTZ
14	F	20	c.1015dupT	p.Asp355*	1	HTZ
15	F	23	c.344G>T	p.Arg115Ile	2	HTZ
16	F	38	c.1633C>T	p.Arg545Cys	1	HTZ
17	M	44	c.2661dup	p.Glu888Argfs*17	1	HTZ
18	M	34	c1646G>A	p.Arg549His	2	HTZ
19	M	44	c.475C>T	p.Arg159*	1	HTZ
20	F	31	c.1553delT	p.Leu518Tyrfs*16	1	HTZ
21	F	18	c.2363G>A	p.Arg788Gln	2	HMZ
22	M	39	c.536+6T>C	p.?	3	HTZ
23	F	44	c.140G>A	p.Arg47Gln	2	HTZ
24	M	23	c.523A>G & c.3269+1G>A	p.Thr175Ala & p.?	1	HTZ & HTZ
25	F	33	c.1712delT	p.Val571Aspfs*16	1	HTZ
26	F	38	c.959C>T	p.Ser320Phe	1	HTZ
27	M	47	c.498C>T & c.2078delC	p.Asp166Asp & p.Pro693Profs*5	1	HTZ
28	M	68	c.140G>A	p.Arg47Gln	2	HMZ
29	F	40	c.3838C>T	p.*1280Arg ext	1	HTZ
30	F	28	c.140G>A	p.Arg47Gln	1	HTZ
31	M	57	c.2324C>T & c.2836G>A	p.Thr775Met & p.Ala946Thr	1	HTZ & HTZ
32	M	29	c.2405G>A	p.Trp802*	1	HTZ
33	F	17	c.140G>A	p.Arg47Gln	2	HTZ
34	M	35	c.1576G>T	p.Val526Phe	1	HTZ
35	F	36	c.959C>T	p.Ser320Phe	1	HTZ
36	F	60	c.578G>A	p.Gly193Glu	2	HTZ
37	M	53	c.202G>A	p.Gly68Arg	1	HTZ

38	F	9	c.1296_1301del	p.Cys433_Gly434del	1	HTZ
39	M	29	c.1327insA	p.Cys433Argfs*7	1	HTZ
40	F	50	c.1896+6T>C	p.?	3	HMZ
41	F	35	c.79A>G	p.Ser27Gly	1	HTZ
42	F	51	c.2800G>A	p.Ala934Thr	1	HTZ
43	F	25	c.217C>G	p.Leu73Val	1	HTZ
44	M	14	c.523A>G	p.Thr175Ala	3	HTZ
45	F	51	c.1005+5G>A	p.?	3	HTZ
46	F	28	c.1778C>T	p.Thr593Met	1	HTZ
47	F	28	c.959C>T	p.Ser320Phe	1	HTZ
48	F	39	c.2800G>A	p.Ala934Thr	1	HTZ
49	F	41	c.3081+5G>C	p.?	3	HTZ
50	M	46	c.1584G>C	p.Glu528Asp	3	HTZ
51	F	48	c.1230+3A>C	p.?	3	HTZ
52	F	44	c.101C>T	p.Thr34Met	1	HTZ
53	F	36	c.1327C>G & c.1769G>A	p.Gln443Glu & p.Arg590Gln	1	HTZ & HTZ
54	F	35	c.2363G>A	p.Arg788Gln	2	HTZ
55	F	62	c.523A>G	p.Thr175Ala	3	HTZ
56	F	42	c.459T>C	p.Phe153Phe	3	HTZ
57	F	47	c.1769G>A	p.Arg590Gln	2	HTZ
58	M	38	c.524C>T	p.Thr175Met	1	HTZ
59	F	24	c.1675C>A	p.Ala559Thr	1	HTZ
60	F	38	c.1974G>A	p.Trp658*	1	HTZ

* 1: disease-causing ; 2: potentially pathogenic ; 3: of unknown significance