Supplemental Table S1. Spectrum of ABCC6 mutations in this patient cohort

ABCC6 Mutation	Mutation type	Clinical Classification	*n* = 138 [%]
c.3421C>T (p.Arg1141*)	Nonsense	pathogenic	49 [35.5]
c.2787+1G>T (p.?)	Splice-site (nonsense)	pathogenic	8 [5.8]
c.3179C>G (p.Pro1060Arg)	Missense	new, pathogenic	4 [2.9]
c.4182delG (p.Lys1394Asnfs*9)	Deletion (nonsense)	pathogenic	4 [2.9]
c.37-1G>A	Splice-site (nonsense)	pathogenic	3 [2.2]
c.3412C>T (p.Arg1138Trp)	Missense	pathogenic	3 [2.2]
c.EX23_EX29del	Deletion (nonsense)	pathogenic	3 [2.2]
c.3883-6G>A (p.?)	Splice-site (missense)	likely pathogenic	3 [2.2]
c.1171A>G (p.Arg391Gly)	Missense	pathogenic	3 [2.2]
c.1553G>A (p.Arg518Glu)	Missense	pathogenic	2 [1.4]
c.2399G>A (p.Gly800Arg)	Missense	new, pathogenic	2 [1.4]
c.2230A>C (p.Thr744Pro)	Missense	new, pathogenic	2 [1.4]
c.346-6G>A (p.?)	Splice-site (missense)	likely benign	2 [1.4]
c.3413C>T (p.Arg1138Glu)	Missense	pathogenic	2 [1.4]
c.3506+2delTAGG (p.?)	Deletion (nonsense)	pathogenic	2 [1.4]
c.3661C>T (p.Arg1221Cys)	Missense	pathogenic	2 [1.4]
c.EX9del (p.*)	Deletion (nonsense)	pathogenic	2 [1.4]
c.EX24_EX27del (p.*)	Deletion (nonsense)	pathogenic	2 [1.4]
c.3088C>T (p.Arg1030*)	Nonsense	pathogenic	2 [1.4]
c.2090C>T (p.Pro697Leu)	Missense	new, pathogenic	2 [1.4]
c.1132C>T (p.Gln378*)	Nonsense	pathogenic	1 [0.7]
c.1460G>A (p.Arg487Gln)	Missense	pathogenic	1 [0.7]
c.1517G>A (p.Trp506*)	Nonsense	pathogenic	1 [0.7]

c.3074T>C (p.Leu1025Pro)	Missense	pathogenic	1 [0.7]
c.1999delG p.(Ala667Glnfs*21)	Deletion (nonsense)	pathogenic	1 [0.7]
c.1589T>C (p.Leu530Pro)	Missense	new, pathogenic	1 [0.7]
c.EX18del (p.?)	Deletion (nonsense)	pathogenic	1 [0.7]
c.3450G>C (p.Asn1150Lys)	Missense	benign	1 [0.7]
c.3903C>T (p.Thr1301Ile)	Missense	pathogenic	1 [0.7]
c.4209-2A>C (p.?)	Splice-site (missense)	likely pathogenic	1 [0.7]
c.EX22del (p.?)	Deletion (nonsense)	pathogenic	1 [0.7]
c.3679_3770insC (p.?)	Insertion (nonsense)	new, likely pathogenic	1 [0.7]
c.3634-3C>A (p.?)	Splice-site (nonsense)	pathogenic	1 [0.7]
c.EX28del (p.?)	Deletion (nonsense)	pathogenic	1 [0.7]
c.2071-1G>A (p.?)	Splice-site (nonsense)	new, pathogenic	1 [0.7]
c.(?_-1)_(*1_?)del (p.0)	Deletion (nonsense)	pathogenic	1 [0.7]
c.2420G>A (p.Arg807Gln)	Missense	pathogenic	1 [0.7]
c.2252T>A (p.Met751Lys)	Missense	pathogenic	1 [0.7]
c.2342C>A (p.Ala781Glu)	Missense	pathogenic	1 [0.7]
c.754C>T (p.Leu252Phe)	Missense	likely pathogenic	1 [0.7]
c.2294G>A (p.Arg765Gln)	Missense	pathogenic	1 [0.7]
c.1987G>A (p.Gly663Ser)	Missense	pathogenic	1 [0.7]
c.2432C>T (p.Thr811Met)	Missense	pathogenic	1 [0.7]
c.3491G>A (p.Arg1164Gln)	Missense	pathogenic	1 [0.7]
c.1091C>G (p.Thr364Arg)	Missense	pathogenic	1 [0.7]
No mutation on second allele detected			11 [8.0]