

S1_Table: Genetic study summary table. Heterozygous *ATP1A3* mutations and protein modifications found in AHC patients in the AHCf registry enrolled from 1997 to 2012.

Nucleotide Δ	Protein Δ	Position on Chromosome 19	Exon	Patient Number	Predicted Consequence
410 C>T	S137F	g.42,490,329	5	1	probably damaging, 1.00
419A>T	Q140L	g.42,490,320	5	1	probably damaging, 0.99
821T>A	I274N		8	4#	probably damaging, 1.00
972G>C	Q324D	g.42,489,093	8	1	probably damaging, 0.99
977T>G	L326R	g.42,489,086	8	1	probably damaging, 0.99
998G>T	C333F	g.42, 486, 254	9	1	probably damaging, 1.00
1786T>C	C596R	g.42,482,323	13	1	probably damaging, 1.00
2263G>A	G755S	g.42,479,781	16	5	probably damaging, 0.99
2264G>T	G755V	g.42,474,694	17	1	probably

					damaging, 1.00
2267G>A	R756H*	g.42,474,691	17	3#	probably damaging, 1.00
2281A>C	N761H	g.42,474,677	17	1	probably damaging, 0.99
2302T>C	Y768H	g.42,474,656	17	1	probably damaging, 0.98
2303A>G	Y768C	g.42,474,655	17	1	probably damaging, 0.99
2305A>C	T769P	g.42,474,653	17	1	probably damaging, 0.99
2314A>C	S772R	g.42,474,644	17	1	possibly damaging, 0.94
2316C>A	S772R	g.42,474,642	17	1	possibly damaging, 0.94
2318A>C	N773H	g.42,474,640	17	1	probably damaging, 0.99
2401G>A	D801N	g.42,474,557	17	58+2*	probably damaging, 0.99
2401G>T	D801Y*	g.42,474,557	17	1	probably damaging, 0.99
2403T>A	D801E	g.42,474,555	17	2*	possibly damaging, 0.79
2411C>T	T804I	g.42,474,547	17	2	probably

					damaging, 0.99
2413G>A	D805N	g.42,474,545	17	1	probably damaging, 0.99
2423C>T	P808L	g.42,474,456	18	1	probably damaging, 0.98
2431T>C	S811P	g.42,474,448	18	3	probably damaging, 0.98
2443G>A	E815K	g.42,474,436	18	38	probably damaging, 1.00
2516T>C	L839P	g.42,474,363	18	1	probably damaging, 0.99
2542+1G>A	splice site	g.42,474,336		1	splice site broken, -29.76%
2542+2T>C	splice site	g.42,474,335		2	splice site broken -29.76%
2702G>C	R901T	g.42,473,054	20	1	probably damaging, 0.99
2751_2753 delTGT	V919del	g.42,473,005- 42,473,003	20	2	deleterious, -8.77%
2780G>A	C927Y	g.42,472,976	20	1	probably damaging, 0.98
2839G>A	G947R	g.42,471,896	21	10	probably damaging, 0.99

2839G>C	G947R	g.42,471,896	21	1	probably damaging, 0.99
2851G>A	E951K	g.42,471, 884	21	1	probably damaging, 0.99

Scoring of predicted consequence was obtained from PolyPhen for missense, Human Splicing Finder for intronic mutations and Provean for deletions. AHC=Alternating Hemiplegia of Childhood; **new, previously unreported mutations in bold**; #multiplex cases; *monozygotic twin pair, **previously reported but with RDP phenotype