H-ABC GENE MUTATION OMNIBUS as at: 7 7 2022

ID	DNA Sequence Change	Amino Acid Change
1	c.4C>T	p.Arg2Trp
2	c.5G>A	p.Arg2Gln
3	c.286G>A	p.Gly96Arg
4	c.293G>A	p.Gly98Asp
5	c.467G>T	p.Arg156Leu
6	c.533C>G	p.Thr178Arg
7	c.533C>T	p.Thr178Met
8	c.535G>C	p.Val179Leu
9	c.538G>A	p.Val180Met
10	c.539T>G	p.Val180Gly
11	c.544C>A	p.Pro182Thr
12	c.568C>T	p.His190Tyr
13	c.716G>A	p.Cys239Tyr
14	c.716G>T	p.Cys239Phe
15	c.730G>A	p.Gly224Ser
16	c.731G>A	p.Gly244Asp
17	c.731G>C	p.Gly244Ala
18	c.731G>T	p.Gly244Val
19	c. 743C>A	p.Ala248Asp
20	c.745G>A	p.Asp249Asn
21	c.755A>T	p.Lys252Met
22	c.763G>A	p.Val255Ile
23	c.785G>A	p.Arg262His
24	c.796T>A	p.Phe266Ile
25	c.845G>C	p.Arg282Pro
26	c.874C>A	p.Gln292Lys
27	c.900G>T	p.Met300Ile
28	c.937G>T	p.Val313Leu
29	c.941C>T	p.Ala314Val
30	c.968T>G	p.Met323Arg
31	c.971A>C	p.Lys324Thr
32	c.1049A>T	p.Lys350Met
33	c.1052C>T	p.Thr351Met
34	c.1054G>A	p.Ala352Thr
35	c.1061G>A	p.Cys354Tyr
36	c.1062C>G	p.Cys354Trp
37	c.1064A>T	p.Asp355Val
38	c.1088T>C	p.Met363Thr
39	c.1091C>A	p.Ala364Asp
40	c.1099T>A	p.Phe367Ile
41	c.1099T>C	p.Phe367Leu
42	c.1162A>G	p.Met388Val
43	c.1163T>C	p.Met388Thr
44	c.1164G>A	p.Met388Ile
45	c.1171C>T	p.Arg391Cys
46	c.1172G>A	p.Arg391His
47	c.1172G>T	p.Arg391Leu
48	c.1181T>G	p.Phe394Cys
49	c.1190G>T	p.Trp397Leu
50	c.1209G>C	p.Met403Ile
51	c.1228G>A	p.Glu410Lys
52	c.1240A>T	p.Asn414Tyr
53	c.1242C>A	p.Asn414Lys

Reference:

1. H-ABC Foundation UK, H-ABC Foundation UK: Gene Mutations Associated with H-ABC Disease (2022).