S2 Table Mutations and phenotypes

Mutations	Total	Men		Women	
		classical	non-classical	classical	non-classical
c.35_47del13	1	1	0	0	0
c.358-6	1	1	0	0	0
c.369+5G>T	1	0	1	0	0
c.370-533_c.1277 del4.5kb	2	0	0	2	0
c.548-1G>A	1	0	0	1	0
c.639+919 G>A	2	0	2	0	0
c.640-2A>C	2	0	0	2	0
c.748_IVS6+8del62	1	1	0	0	0
c.802-3_802-2delCA	1	1	0	0	0
c.1000-2 A>G	2	1	0	1	0
duplication exon 3+4	1	1	0	0	0
exon 1 deletion	3	2	0	1	0
IVS2+1G>A	1	1	0	0	0
IVS3+1G>A	4	3	0	0	1
IVS4+1G>A	1	0	0	1	0
IVS5-3_2delCA	1	0	0	1	0
IVS6-10G>A	1	1	0	0	0
IVS6-2A>T	1	0	0	1	0
p.Ala135Val	2	1	0	1	0
p.Ala143Pro	26	7	0	19	0
p.Ala143Thr	2	1	0	1	0
p.Ala156ProFS*9	2	1	0	0	1
p.Ala156Thr	1	0	0	1	0
p.Ala257Ser	1	1	0	0	0
p.Ala285Asp	5	0	2	3	0
p.Ala309Pro	2	0	0	1	1
p.Ala31Val	1	0	0	1	0
p.Ala348Pro	2	1	0	1	0
p.Arg112Cys	5	5	0	0	0
p.Arg112His	4	0	3	0	1
p.Arg220*	10	5	0	5	0
p.Arg227*	18	8	0	8	2
p.Arg227Gln	2	2	0	0	0
p.Arg301*	4	2	0	2	0
p.Arg301Gln	4	0	2	0	2
p.Arg301Pro	1	1	0	0	0
p.Arg332Lys Fs*7	4	2	0	2	0
p.Arg342*	8	4	0	4	0
p.Arg342Gln	13	6	0	7	0
p.Arg342Leu	4	2	0	2	0
p.Arg363Cys	1	0	1	0	0
p.Arg49Cys	1	1	0	0	0
p.Arg49Leu	2	2	0	0	0
p.Asn215Ser	49	2	33	0	14
p.Asn224Gly	1	0	0	1	0
p.Asn298Ser	1	0	1	0	0
p.Asn33Asp	1	1	0	0	0
p.Asn34Lys fs*22	1	0	0	1	0
p.Asn355_Ile359del	1	0	0	1	0

p.Asn408Ile fs*10	4	1	0	1	2
p.Asn53Leu fs*57	2	1	0	1	0
p.Asp136Glu	2	1	0	0	1
p.Asp136Tyr	7	4	0	2	1
p.Asp165Val	1	1	0	0	0
p.Asp170Asn	1	1	0	0	0
p.Asp234del	1	0	0	1	0
p.Asp299Glu	1	0	1	0	0
p.Asp55Thr fs*66	1	1	0	0	0
p.Cys174Val fs*4	6	1	0	5	0
p.Cys202Trp	1	0	0	1	0
p.Cys63Ala	1	1	0	0	0
p.Gin386*	3	0	0	3	0
p.Gln107*	1	0	0	0	1
p.Gln280His	2	1	0	0	1
p.Gln312*	1	0	0	1	0
p.Gln321_Asp322 DelInsHisAsn	1	1	0	0	0
p.Gln321Leu	2	0	1	1	0
p.Gln386*	2	1	0	1	0
p.Gln416*	2	1	0	0	1
p.Glu338Lys	3	0	0	1	2
p.Glu341Lys	2	2	0	0	0
p.Glu358Asp fs*16	3	0	0	2	1
p.Gly132Glu	1	1	0	0	0
p.Gly183Asp	2	1	0	0	1
p.Gly260Glu	1	1	0	0	0
p.Gly261Val	1	1	0	0	0
p.Gly325Ala fs*23	2	1	0	1	0
p.Gly325Ser	2	0	1	0	1
p.Gly35Arg	2	1	0	0	1
p.Gly361Arg	4	3	0	1	0
p.Gly373Asp	2	1	0	1	0
p.Gly375Glu fs	2	2	0	0	0
p.Gly43Val	1	0	0	1	0
p.Ile232Thr	1	0	1	0	0
p.Ile253Leu fs*16	1	1	0	0	0
p.Ile317Thr	7	2	0	3	2
p.Ile319Leu fs*10	3	2	0	1	0
p.Ile319Thr	1	0	1	0	0
p.Ile91Thr	1	0	0	0	1
p.Leu129Pro	5	3	0	1	1
p.Leu166Pro	1	0	1	0	0
p.Leu166Ser	1	0	0	1	0
p.Leu21Pro	2	0	0	1	1
p.Leu268Ser	1	1	0	0	0
p.Leu311Val	1	1	0	0	0
p.Leu347Phe fs*28	2	0	0	1	1
p.Leu372Pro	2	1	0	1	0
p.Leu403*	2	2	0	0	0
p.Leu414Ser	1	0	0	1	0
p.Leu54Pro	1	0	1	0	0
p.Lys240Gly FS*9	4	2	0	2	0
p.Met187Ser FS*5	1	1	0	0	0
p.Met187Val	1	0	0	0	1
p.Met1Thr	2	2	0	0	0
p.Met42Val	2	1	0	1	0

p.Met72Arg	1	1	0	0	0
p.Phe169Ser	1	0	0	1	0
p.Phe18Ser	5	0	0	4	1
p.Pro205Thr	10	3	0	5	2
p.Pro293His	1	0	0	0	1
p.Pro293Leu	2	1	0	0	1
p.Pro389Ala	4	0	2	0	2
p.Pro409Thr	2	0	0	0	2
p.Ser126Gly	1	0	0	0	1
p.Ser345Arg fs*29	1	1	0	0	0
p.Ser345Pro	6	1	1	4	0
p.Thr282Ile	2	2	0	0	0
p.Thr410Ile	2	0	0	2	0
p.Trp204*	1	1	0	0	0
p.Trp204Cys	1	0	0	0	1
p.Trp209*	1	1	0	0	0
p.Trp226*	3	2	0	1	0
p.Trp236Cys	2	1	0	1	0
p.Trp236Leu	1	0	0	1	0
p.Trp277*	1	1	0	0	0
p.Trp349*	5	2	0	2	1
p.Trp399*	2	2	0	0	0
p.Trp47Gly	1	0	0	1	0
p.Trp81Ser	1	1	0	0	0
p.Tyr134fs*1	1	0	0	1	0
p.Tyr134Ser	1	0	0	1	0
p.Tyr184*	2	0	0	2	0
p.Tyr207fs	1	0	0	1	0
p.Val269Ala	3	1	0	2	0
p.Val316Glu	2	1	0	0	1