

TDD VARIANTS

VARIANT	EXON #	HGVS PROTEIN CHANGE	EXPECTED CHANGE*
22q11.21 del	-	-	-
Exon 1-2 del	-	-	-
Exon 2-8 del	-	-	-
Exon 3-5 del	-	-	-
Exon 3-9 del	-	-	-
Exon 4-6 del	-	-	-
Exon 4-9 del	-	-	-
Exon 5-9 del	-	-	-
Exon 6 del	-	-	-
c.4delT	2	p.Cys2Alafs*35	Frameshift
c.6_9delCATC	2	p.Ile3=fs*32	Frameshift
c.11_13delTCT	2	p.Phe5del	Inframe deletion
c15_17delCTT	2	p.Phe6del	Inframe deletion
c.35_36CT	2	p.Pro12Argfs*16	Frameshift
c.57-1G>C	-	-	Intronic
c.59T>G	3	p.Leu20Pro	Missense
c.77G>A	3	p.Arg26Lys	Missense
c.94C>T	3	p.Glu32*	Stop gained
c.95G>A	3	p.Arg23Gln	Missense
c.119G>A	4	p.Trp40*	Stop gained
c.120G>A	4	p.Trp40*	Stop gained
c149T>C	4	p.Leu50Pro	Missense

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c.217C>T	4	p.Arg73*	Stop gained
c.220A>C	4	p.Thr74Pro	Missense
c.256C>T	4	p.Arg86*	Stop gained
c.262C>T	4	p.Arg88*	Stop gained
c.265G>T	4	p.Gly89Cys	Missense
c.280delC	5	p.His94Thrfs*3	Frameshift
c.336delG	5	p.Glu112=fs*10	Frameshift
c.338delG	5	p.Gly113Alafs*10	Frameshift
c.353G>T	5	p.Gly118Val	Missense
c.380+1 G>A	-	-	Intronic
c.385C>T	6	p.Arg129*	Stop gained
c.418C>T	6	p.Arg140*	Stop gained
c.420_421delGA	6	p.Glu142Alafs*2	Frameshift
c.451+2T>A	-	-	Intronic
c.460G>A	7	p.Gly154Arg	Missense
c.544C>T	7	p.Gln182*	Stop gained
c.569_592del	7	p.Ile190_Leu197del	Inframe deletion
c.605+1G>A	-	-	Intronic
c.707C>T	8	p.Thr236Ile	Missense
c.711-3C>G	-	-	Intronic
c.728+1G>A	-	-	Intronic

Figure 1. Current TDD Variants. Adapted from DOI: 10.1002/ajmg.a.62543, DOI: <https://doi.org/10.1016/j.gim.2022.11.020>, and DOI:10.1016/j.hrthm.2022.05.009
 *Definitions for terms used in this chart can be found in the Glossary