

**Table 2:** Recurrent mutations in the exon 24-32 region and their type of presentation

<b>Mutation</b>	<b>Effect</b>	<b>Number of event</b>	<b>Type of presentation</b>	<b>References</b>
c.2860C>T	p.Arg954Cys	2	Classical MFS (2)	35; L. Adès, pers. comm.
c.2953G>A	p.Gly985Arg	2	Classical MFS (1), probable MFS (1)	36;37
c.2980G>T	p.Glu994X	2	Classical MFS (2)	35;38
c.3037G>A	p.Gly1013Arg	7	Neonatal MFS (1), severe MFS (3), classical MFS (2), probable MFS (1)	8;35;39-42
c.3058A>G	p.Thr1020Ala	2	Probable MFS (1), other type I fibrillinopathy (1)	8; C. Boileau, pers. comm.
c.3095G>A	p.Cys1032Tyr	2	Neonatal MFS (2)	35;43
c.3143T>C	p.Ile1048Thr	5	Neonatal MFS (3), severe MFS (1), probable MFS (1)	11;35;44-45
c.3157T>C	p.Cys1053Arg	2	Neonatal MFS (2)	7;11
c.3202T>C	p.Cys1068Arg	2	Neonatal MFS (1), probable MFS (1)	L. Adès, pers.comm. ; P. Robinson, pers. comm
c.3217G>A	p.Glu1073Lys	3	Neonatal MFS (2), probable MFS (1)	7;39;46
c.3299G>T	p.Gly1100Val	2	Classical MFS (2)	46; C. Boileau, pers.comm.
c.3302A>G	p.Tyr1101Cys	6	Classical MFS (5), probable MFS (1)	36;42;47-48; P. Robinson, pers.comm. ; C. Boileau, pers.comm.
c.3344A>G	p.Asp1115Gly	2	Classical MFS (1), other type I fibrillinopathy (1)	8; P. Robinson, pers.comm.
c.3350G>A	p.Cys1117Tyr	2	Severe MFS (1), classical MFS (1)	42;49
c.3373C>T	p.Arg1125X	3	Probable MFS (1), other type I fibrillinopathy (2)	37; A. De Paepe, pers.comm. ; J.C. Hyland, pers.comm.
c.3388delC	p.His1130IlefsX32	4	Classical MFS (4)	11;46
c.3410G>C	p.Arg1137Pro	2	Classical MFS (2)	50
c.3412T>C	p.Cys1138Arg	2	Severe MFS (1), classical MFS (1)	11; De Paepe, pers. comm.
c.3463G>A	p.Asp1155Asn	2	Classical MFS (1), other type I fibrillinopathy (1)	42;51
c.3511T>C	p.Cys1171Arg	2	Classical MFS (1), probable MFS (1)	31; L. Adès, pers. comm.
c.3656A>G	p.Tyr1219Cys	2	Classical MFS (2)	48
c.3668G>A	p.Cys1223Tyr	2	Severe MFS (1), classical MFS (1)	52;53
c.3725G>A	p.Cys1242Tyr	3	Severe MFS (1), classical MFS (1), probable MFS (1)	6;31; P. Robinson, pers. comm.
c.3976T>C	p.Cys1326Arg	2	Neonatal MFS (1), severe MFS (1)	31;35

Pers. comm. : personal communication