APPENDIX 2**. Summary of clinical Findings and Mutations in Patients with *FBN1* and *ADAMTS10* Mutations**

Exon	Patient ID	Gene	Variations			State	Bioinformatic analysis		age (years)	Other organs		Reported phenotype	Diagnosis in this study
			Nucleotide	Amino acid	Domain		P/SS	SIFT		SS	CS		
2	QT448	*FBN1*	c.184C>T	p.Arg62Cys	NH2 Region	Het	PrD	D	2	—	NA	MFS[30]	MFS
2	QT711	*FBN1*	c.184C>T	p.Arg62Cys	NH2 Region	Het	PrD	D	2	—	NA	MFS[30]	MFS
2	QT1244	*FBN1*	c.184C>T	p.Arg62Cys	NH2 Region	Het	PrD	D	3	m	—*	MFS[30]	MFS
2	QT1279	*FBN1*	c.184C>T	p.Arg62Cys	NH2 Region	Het	PrD	D	16	—	NA	MFS[30]	MFS
2	QT925	*FBN1*	c.188A>G	p.Tyr63Cys	Cys-rich	Het	PrD	D	3	—	—*	EL[31]	IEL
4	QT1065	*FBN1*	c.364C>T	p.Arg122Cys	EGF-like#02	Het	PrD	T	6	m	—	MFS[32]	MFS
4	QT1268	*FBN1*	c.364C>T	p.Arg122Cys	EGF-like#02	Het	PrD	T	9	—	NA	MFS[32]	MFS
4	QT1293	*FBN1*	c.364C>T	p.Arg122Cys	EGF-like#02	Het	PrD	T	3	m	—	MFS[32]	MFS
6	QT1346	*FBN1*	c.649T>G	p.Trp217Gly	TB 1	Het	PrD	D	3	—	—	MFS[33]	MFS
6	QT1296	*FBN1*	c.718C>T	p.Arg240Cys	Hybrid#01	Het	PrD	T	9	m	—	MFS[32]	MFS
11	QT581	*FBN1*	c.1463G>T	p.Cys488Phe	EGF-like#6	Het	PrD	D	10	m	NA	MFS[34]	MFS
12	QT1280	*FBN1*	c.1556A>G	p.Tyr519Cys	cbEGF-like#7	Het	PrD	D	2	m	—	MFS[34]	MFS
13	QT444	*FBN1*	c.1601G>A	p.Cys534Tyr	cbEGF-like#8	Het	PrD	D	8	m	NA	MFS[35]	MFS
13	QT243	*FBN1*	c.1633C>T	p.Arg545Cys	cbEGF-like#8	Het	PrD	T	2	—	NA	MFS[36]	MFS
13	QT449	*FBN1*	c.1633C>T	p.Arg545Cys	cbEGF-like#8	Het	PrD	T	3	—	NA	MFS[36]	MFS
15	QT723	*FBN1*	c.1916G>A	p.Cys639Tyr	cbEGF-like#10	Het	PrD	D	5	—	—	NA[37]	IEL
15	QT1345	*FBN1*	c.1916G>A	p.Cys639Tyr	cbEGF-like#10	Het	PrD	D	5	—	—	NA[37]	IEL
15	QT1252	*FBN1*	c.1955G>T	p.Cys652Phe	cbEGF-like#10	Het	PrD	D	16	m	—*	novel	IMFS
18	QT1228	*FBN1*	c.2222delA	p.Asn741Thrfs*31	cbEGF-like#11	Het	NA	NA	15	m	m	novel	MFS
19	QT156	*FBN1*	c.2306G>A	p.Cys769Tyr	cbEGF-like#12	Het	PrD	D	5	NA	NA	MFS[38]	MFS
19	QT1141	*FBN1*	c.2413T>C	p.Cys805Arg	cbEGF-like#12	Het	PrD	D	12	m	—	AN[23]	MFS

20	QT150	*FBN1*	c.2433C>G	p.Cys811Trp	cbEGF-like#13	Het	PrD	D	2	m	NA	MFS[34]	MFS
22	HM854	*FBN1*	c.2722T>C	p.Cys908Arg	Hybrid #02	Het	PrD	D	2	m	—*	MFS[39]	MFS
22	QT1142	*FBN1*	c.2728+3A>G	not tested	cbEGF-like#14	Het	SSA		15	m	—	MFS[40]	MFS
24	QT229	*FBN1*	c.2920C>T	p.Arg974Cys	TB 5	Het	PrD	T	33	NA	NA	MFS[31]	MFS
25	QT905	*FBN1*	c.3083A>G	p.Asp1028Gly	cbEGF-like#15	Het	PrD	D	10	m	—	IMFS[31]	IMFS
35	QT753	*FBN1*	c.4381T>C	p.Cys1461Arg	cbEGF-like#25	Het	PrD	D	8	m	—	novel	MFS
37	QT394	*FBN1*	c.4588C>T	p.Arg1530Cys	8-Cys #04	Het	PrD	T	23	m	M*	MFS[41]	MFS
39	QT228	*FBN1*	c.4898G>C	p.Cys1633Ser	cbEGF-like#27	Het	PrD	D	5	m	M	IMFS[31]	MFS
44	QT1026	*FBN1*	c.5504G>A	p.Cys1835Tyr	cbEGF-like#30	Het	PrD	D	4	m	NA	MFS[42]	MFS
46	QT126	*FBN1*	c.5788+5G>A	not tested	cbEGF-like#32	Het	SSA		7	m	M	MFS[43]	MFS
46	QT459	*FBN1*	c.5788+5G>A	not tested	cbEGF-like#32	Het	SSA		4	m	M	MFS[43]	MFS
46	QT755	*FBN1*	c.5788+5G>A	not tested	cbEGF-like#32	Het	SSA		5	m	NA	MFS[43]	MFS
60	QT622	*FBN1*	c.7559C>T	p.Thr2520Met	cbEGF-like#43	Het	PrD	T	13	—	m	MFS[31]	MFS
11	QT401	*ADAMT-*	c.1586G>A	p.Gly529Glu	Disintegrin	Com	PrD	D	7	△m	▲m	novel	WMS
19		*S10*	c.2485T>A	p.Trp829Arg	TSP type-1 2	Het	PrD	T					

Note: Het=heterozygous; P/SS=polyphen-2/splice site prediction; PrD=probably damaging; SSA=splicing site abolished; D=damaging; T=tolerated; SS=skeletal system; CS=cardiovascular system; M=major involvement; m=minor involvement; "—"=not involved. NA=not done or not available. *=signifies affected parent; IEL=isolated EL; IMFS=incomplete MFS; AN=aneurysm. ∆m= short status, brachydactyly, joint stiffness, dental abnormalities; ▲m= had ventricular septal defect repair operation at five years old.