Supplementary

Table S1 Summary of gene mutations in MFS patients diagnosed using revised Ghent criteria

ID	Diagnosis of type	Gene	Variants	Mutation type	Exon	State	First reported	Domain	SIFT	Polyphen2	Mutation taster
F1	AO + Gene	*FBN1*	c.6049T>C	Missense	49	Het	15733436	cb EGF-like #31	D	D	D
F2	AO + Gene	*FBN1*	c.640G>A	Missense	6	Het	15733436	hybrid module #01	D	D	D
F3	AO + Gene	*FBN1*	c.2762G>A	Missense	23	Het	25652356	cb EGF-like #10	D	D	D
F4	AO + Gene	*FBN1*	c.4204T>C	Missense	33	Het	16222657	cb EGF-like #19	D	D	D
F5	AO + Gene	*FBN1*	c.1633C>T	Missense	13	Het	9338581	cb EGF-like #04	D	D	D
F6	AO + Gene	*FBN1*	c.478T>C	Missense	5	Het	17657824	EGF-like #03	D	D	D
F7	AO + Gene	*FBN1*	c.2369G>C	Missense	19	Het	NA	cb EGF-like #08	D	D	D
F8	AO + Gene	*FBN1*	c.364C>T	Missense	4	Het	8040326	EGF-like #02	D	D	A
F9	AO + Gene	*FBN1*	c.5788 + 5G>A	Splicing	46-47	Het	7611299	cb EGF-like #29	/	/	/
F10	AO + Gene	*FBN1*	c.367T>C	Missense	4	Het	16222657	EGF-like #02	D	D	D
F11	AO + Gene	*FBN1*	c.1879C>T	Missense	15	Het	8004112	cb EGF-like #06	D	D	D
F12	AO + Gene	*FBN1*	c.1879C>T	Missense	15	Het	8004112	cb EGF-like #06	D	D	D
F13	AO + Gene	*FBN1*	c.2179T>C	Missense	18	Het	NA (awaiting report)	cb EGF-like #07	D	D	D
F14	AO + Gene	*FBN1*	c.2496T>G	Missense	20	Het	NA (awaiting report)	cb EGF-like #09	D	D	D
F15	AO + Gene	*FBN1*	c.3346G>C	Missense	27	Het	NA (awaiting report)	cb EGF-like #13	D	D	D
F16	AO + Gene	*FBN1*	c.4096G>A	Missense	33	Het	14695540	cb EGF-like #19	D	D	D
F17	AO + Gene	*FBN1*	c.3872G>A	Missense	31	Het	NA	cb EGF-like #17	D	D	D
F18	AO + Gene	*FBN1*	c.2471G>T	Missense	23	Het	NA	cb EGF-like #10	D	D	D
F19	AO + Gene	*FBN1*	c.1633C>T	Missense	13	Het	9338581	cb EGF-like #04	D	D	D
F20	AO + Gene	*FBN1*	c.1633C>T	Missense	13	Het	9338581	cb EGF-like #04	D	D	D
F21	AO + Gene	*FBN1*	c.4260C>G	Missense	34	Het	19293843	cb EGF-like #20	D	D	D
F22	AO + Gene	*FBN1*	c.6662G>A	Missense	54	Het	26787436	cb EGF-like #34	D	D	D
F23	AO + Gene	*FBN1*	c.7936T>C	Missense	63	Het	21542060	cb EGF-like #42	D	D	D
F24	AO + Gene	*FBN1*	c.2168-2A>G	Splicing	17-18	Het	12203992	cb EGF-like #07	/	/	/
F25	AO + Gene	*FBN1*	c.4460-8G>A	Splicing	35-36	Het	11700157	cb EGF-like #22	/	/	/
F26	AO + Gene	*FBN1*	c.4816 + 2dup	Splicing	38-39	Het	NA	cb EGF-like #23	/	/	/
F27	AO + Gene	*FBN1*	c.5066-2A>G	Splicing	40-41	Het	16222657	TGFBF #05	/	/	/
F28	AO + Gene	*FBN1*	c.6932G>A	Missense	56	Com Het	NA	cb EGF-like #36	T	P	N
	AO + Gene	*FBN1*	c.2723G>A	Missense	22		11170092	hybrid motif #02	D	D	D
F29	AO + Gene	*FBN1*	c.1714 + 2T>G	Splicing	13-14	Com Het	NA	cb EGF-like #05	/	/	D
	AO + Gene	*FBN1*	c.1711delG	Frameshift	13		NA	cb EGF-like #04	/	/	/
F30	AO + Gene	*FBN1*	c.1291C>T	Missense	10	Het	NA	proline-rich	T	P	D
F31	AO + Gene	*FBN1*	c.7754T>C	Missense	62	Het	10464652	cb EGF-like #41	T	P	D
F32	AO + Gene	*FBN1*	c.4891T>C	Missense	39	Het	NA	cb EGF-like #23	D	D	D
F33	AO + Gene	*FBN1*	c.3920G>A	Missense	32	Het	17657824	cb EGF-like #17	D	D	D
F34	AO + EL	/									
F35	AO + EL	/									
F36	AO + EL	/									
F37	AO + EL	/									
F38	AO + EL	/									
F39	AO + EL	/									
F40	AO + EL	/									
F41	AO + EL	/									
F42	AO + EL	/									
F43	AO + EL	/									
F44	AO + EL	/									
F45	AO + EL	/									
F46	AO + EL	/									
F47	AO + EL	/									
F48	AO + EL	/									
F49	AO + EL	/									
F50	AO + EL	/									
F51	AO + EL	/									
F52	AO + EL	/									

SIFT: D: damaging, T: tolerated; polyphen2 D: probably damaging, P, possibly damaging; mutation taster D: disease causing, A: disease causing automatic, N: polymorphism; "/": not available. MFS, Marfan syndrome; Het, heterozygous; CNV, copy number variants; AO, aortic diameter; EL, ectopia lentis.