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.4086G>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   | NA            | cb EGF-like #36  | T    | P         | N              |
| F29 | AO + Gene          | FBN1  | c.2732G>A | Missense       | 22   | Het   | 11170092      | hybrid motif #02 | D    | D         | D              |
| F30 | AO + Gene          | FBN1  | c.1714 + 2T>G | Splicing | 13-14 | Com | NA | cb EGF-like #05 | /    | /         | D              |
| F31 | AO + Gene          | FBN1  | c.1711delG | Frameshift | 13   | Het | NA | cb EGF-like #04 | /    | /         | /              |
| F32 | AO + Gene          | FBN1  | c.1291C>T | Missense       | 10   | Het   | NA            | proline-rich    | T    | P         | D              |
| F33 | AO + Gene          | FBN1  | c.7754T>C | Missense       | 62   | Het   | 10464652      | cb EGF-like #41  | T    | P         | 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.