

Appendix 2. Clinical details of the patients II:2, III:12, and family member IV:4.

Individual ID (as indicated in Figure 1)	II:2	III:12	IV:4
Age (Years)	66	32	2
Sex	F	F	F
Ocular system			
Ectopia lentis	+IOL	+	-
Myopia	+	+	-
Abnormally flat cornea	-	-	-
Early development of nuclear cataract	-	-	-
Strabismus	-	+	-
Hypoplastic iris or ciliary muscle	-	-	-
Glaucoma	-	-	-
Retinal detachment	-	-	-
Cardiovascular system			
Aortic root dimension (mm)	N	34.0	N
Mitral valve prolapse	-	+mild	-
Dilation of pulmonary artery	-	-	-
Dilation or dissection of descending thoracic or abdominal aorta	-	-	-
Skeletal system			
Height (H; cm)	168	175	100
Arm span (AS; cm)	178	174	101
AS/H (normal <1.05)	1.06	0.99	0.99
Pectus carinatum	-	+	-
Pectus excavatum	-	-	-
Scoliosis	-	-	-
Arachnodactyly	+	+	-
High palate with dental crowding	-	-	-
Joint hypermobility	+	+	-
Characteristic face (long narrow head, hollow cheeks, hollow-eyed, recessed or protruding jaw)	-	+	-
Flat feet	-	-	-

Other manifestations			
Hyperextensible skin	-	-	-
Striae	-	-	-
Hernia	-	-	-

Each patient in the family has bilateral ectopia lentis, myopia, arachnodactyly, and joint hypermobility. Only the proband displays abnormalities of the cardiovascular system. Individual IV:4 appears normal. Abbreviations: M: male; F: female; IOL: intraocular lens; AS/H: arm span/height ratio; +: present; -: absent. N: normal.