

### APPENDIX 3. Clinical Features of the five Probands without Mutation

Patient ID	Age	SS	OS	CS	Additional information	Diagnosis
QT24	30	m	EL	—	subject's brother was diagnosed with MFS	MFS
QT539	3	m	EL	NA	subject's father was diagnosed with MFS	MFS
QT1149	2	wrist and thumb signs; scoliosis	EL	AD	hypoplasia iris	MFS
QT1226	3	pectus carinatum; arachnodactyly	EL	AD	—	MFS
LE2527	6	lanky stature; arachnodactyly	EL	AD	subject's father died of aortic ruptures	MFS

Note: SS=skeletal system; OS=ocular system; CS=cardiovascular system; “—”=not involved; NA=not done or not available; EL= ectopia lentis ;

AD= aorta dilatation.