ANALYTE:
FBN1

NAME:	fibrillin 1
SYMBOL:	FBN1
VERSION OF ORPHANET:	2023-06-22 14:14:43
SYNONYMS:	MASS Marfan syndrome OCTD SGS asprosin
XREF(S):	Orphanet Ensembl Genatlas HGNC OMIM Reactome SwissProt
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RELATED CONTENT

Related Genetic Tests

- Aneurysm, Thoracic Aortic, familial (gene panel)
- Cataract (gene panel)
- Congenital malformation (gene panel - 1721 genes)
- Congenital malformation gene panel
- Congenital structural heart defects (gene panel)
- Dermatogenetic panel, severe, rare and hereditary genodermatoses (gene panel - 394 genes)
- Ectopia lentis
- Familial Thoracic Aortic Aneurysm (gene panel)
- Glaucoma (gene panel)
- Heart / Cardio disorders / Cardiopathy (gene panel)
- Intellectual disability (virtual gene panel)
- Marfan Syndrome
- Myopia (early onset high myopia)
- Neurodevelopmental disorders (1300 genes)
- Neurodevelopmental disorders gene panel
- Skeletal dysplasia (gene panel)
- Skeletal dysplasia (gene panel)
- Skeletal dysplasia (gene panel)
- Stroke (gene panel)
- Weill-Marchesani syndrome

Related Diseases

- Acromicric dysplasia

- Familial thoracic aortic aneurysm and aortic dissection
- Geleophysic dysplasia
- Glaucoma-ectopia lentis-microspherophakia-stiff joints-short stature syndrome
- Isolated ectopia lentis
- Marfan syndrome type 1
- Neonatal Marfan syndrome
- Progeroid and marfanoid aspect-lipodystrophy syndrome
- Shprintzen-Goldberg syndrome
- Stiff skin syndrome
- Weill-Marchesani syndrome

Related Gene Panels

- Cataract - UGent
- Congenital malformation (1721 genes) - ULB
- Congenital malformation gene panel - VUB
- Congenital structural heart defects - UGent
- Dermatogenetic / severe, rare and hereditary genodermatoses (394 genes) - ULB
- Ectopia lentis (3 gènes)
- Familial Thoracic Aortic Aneurysm (21 genes) - UGent
- Familial Thoracic Aortic Aneurysm (genepanel) - UZA
- Familiale thoracale aorta aneurysmata (19 genes) - UGent
- Glaucoma - UGent
- Intellectual disability (gene panel)
- Myopia gene panel - UGent
- Neurodevelopmental disorders (1300 genes) - ULB
- Neurodevelopmental disorders: developmental delay, intellectual disability, autistic disorders (1162 genes) - VUB
- Skeletal dysplasia (394 genes) - VUB
- Skeletal dysplasia (genepanel) - UZA
- Skeletal dysplasia - UGent
- Stroke - UGent
- Weill-Marchesani - UGent
- cardiopathy panel - UGent
