DISEASE:
Shprintzen-Goldberg syndrome

NAME:	Shprintzen-Goldberg syndrome
DESCRIPTION:	A rare genetic disorder characterized by craniosynostosis, craniofacial and skeletal abnormalities, marfanoid habitus, cardiac anomalies, neurological abnormalities, and intellectual disability.
ORPHACODE:	2462
SYNONYMS:	Marfanoid craniosynostosis syndrome SGS
XREF(S):	Orphanet ICD-10 OMIM MeSH
ANALYTE(S):	SKI FBN1
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Genetic Tests

- Aneurysm, Thoracic Aortic, familial (gene panel)
- Craniosynostosis (gene panel)
- Familial Thoracic Aortic Aneurysm (gene panel)
- cleft lip with/whitout cleft palate (virtual gene panel)

Related Laboratories

- Centre de Génétique Médicale UCL
- Centrum Medische Genetica - UZ Antwerpen
- Centrum Medische Genetica - UZ Gent
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- fibrillin 1
- SKI proto-oncogene

Related Gene Panels

- Cleft lip and palate / dysmorphic facial features / craniofacial anomalies (255 genes)) - UCL
- Craniosynostosis (32 genes) - KUL
- Familial Thoracic Aortic Aneurysm (21 genes) - UGent

- Familial Thoracic Aortic Aneurysm (genepanel) - UZA
