DISEASE:
Myhre syndrome

NAME:	Myhre syndrome
DESCRIPTION:	A rare multiple congenital anomalies syndrome characterized by short stature, distinctive facial dysmorphism, brachydactyly, stiff and thick skin, muscular pseudohypertrophy, restricted joint mobility, hearing loss, and variable intellectual disability. Cardiovascular and respiratory involvement are common.
ORPHACODE:	2588
SYNONYMS:	Facial dysmorphism-intellectual disability-short stature-deafness syndrome Facial dysmorphism-intellectual disability-short stature-hearing loss syndrome
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	SMAD4
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RELATED CONTENT

Related Genetic Tests

- Myhre syndrome (hot spot mutation - p.I500)
- Rendu-Osler-Weber disease (4 genes)

Related Laboratories

- Centre de Génétique Médicale UCL
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)

Related Analytes

- SMAD family member 4

Related Gene Panels

- Rendu/Osler/weber (4 genes) - UCL