DISEASE:
Sheldon-Hall syndrome

NAME:	Sheldon-Hall syndrome
DESCRIPTION:	Sheldon-Hall syndrome (SHS) is a rare multiple congenital contracture syndrome characterized by contractures of the distal joints of the limbs, triangular face, downslanting palpebral fissures, small mouth, and high arched palate.
ORPHACODE:	1147
SYNONYMS:	Distal arthrogryposis type 2B Freeman-Sheldon syndrome variant
XREF(S):	Orphanet OMIM OMIM OMIM ICD-10
ANALYTE(S):	NALCN TNNI2 TNNT3 TPM2 MYH3
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- Neuromuscular disorders : congenital & distal myopathy, congenital muscle dystrophy / Limb-girdle muscular dystrophy / Rhabdomyolysis / Myopathy (with prominent contractures) / distal artrogryposis (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- myosin heavy chain 3
- sodium leak channel, non-selective
- troponin I2, fast skeletal type
- troponin T3, fast skeletal type
- tropomyosin 2

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