DISEASE:
Distal arthrogryposis type 5D

NAME:	Distal arthrogryposis type 5D
DESCRIPTION:	Distal arthrogryposis type 5D is a rare subtype of distal arthrogryposis syndrome characterized by arthrogryposis multiplex congenita affecting the hands, feet, ankle, shoulders and/or neck, with camptodactyly of the fingers and limited knee and hip extension, associated with asymmetric ptosis and, less frequently, other ocular manifestations (e.g. ophthalmoplegia, strabismus). Affected individuals frequently have a bulbous nose, furrowed tongue, micro/retrognathia, a short neck, congenital hip dislocation, club feet, scoliosis and short stature.
ORPHACODE:	329457
SYNONYMS:	DA5D Distal arthrogryposis type 5 without ophthalmoparesis Distal arthrogryposis type 5 without ophthalmoplegia
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	ECEL1
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RELATED CONTENT

Related Genetic Tests

- 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

- endothelin converting enzyme like 1

Related Gene Panels

- Neuromuscular disorders (166 genes) - VUB