DISEASE:
Congenital muscular dystrophy, Ullrich type

NAME:	Congenital muscular dystrophy, Ullrich type
DESCRIPTION:	Ullrich congenital muscular dystrophy (UCMD) is characterized by early-onset, generalized and slowly progressive muscle weakness, multiple proximal joint contractures, marked hypermobility of the distal joints and normal intelligence.
ORPHACODE:	75840
SYNONYMS:	Scleroatonic muscular dystrophy UCMD Ullrich disease
XREF(S):	Orphanet ICD-10 OMIM OMIM
ANALYTE(S):	COL6A1 COL6A2 COL6A3 COL12A1
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RELATED CONTENT

Related Genetic Tests

- Bethlem myopathy / Ullrich congenital muscular dystrophy / Myosclerosis Myopathy
- Ehlers-Danlos syndroom, EDS (gene panel)
- 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
- Centrum Medische Genetica - UZ Gent

Related Analytes

- collagen type XII alpha 1 chain
- collagen type VI alpha 1 chain
- collagen type VI alpha 2 chain
- collagen type VI alpha 3 chain

Related Gene Panels

- Bethlem myopathy / Ullrich / Myosclerosis Myopathy - UGent
- Ehlers-Danlos syndrome -UGent
- Neuromuscular disorders (166 genes) - VUB
