DISEASE:
Primary dystonia, DYT27 type

NAME:	Primary dystonia, DYT27 type
DESCRIPTION:	A rare genetic dystonia characterized by focal or segmental isolated dystonia involving the face, neck, upper limbs (commonly writing dystonia), larynx, or trunk, with an onset from childhood to early adulthood. Dystonia may be tremulous, giving rise to head or hand tremor. Mode of inheritance is autosomal recessive.
ORPHACODE:	464440
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	COL6A3
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RELATED CONTENT

Related Genetic Tests

- Bethlem myopathy / Ullrich congenital muscular dystrophy / Myosclerosis Myopathy
- Dystonia (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
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- collagen type VI alpha 3 chain

Related Gene Panels

- Bethlem myopathy / Ullrich / Myosclerosis Myopathy - UGent
- Dystonia (68 genes) - KUL
- Neuromuscular disorders (166 genes) - VUB