DISEASE:
Musculocontractural Ehlers-Danlos syndrome

NAME:	Musculocontractural Ehlers-Danlos syndrome
DESCRIPTION:	A rare systemic disease characterized by congenital multiple contractures, characteristic craniofacial features (like large fontanel, hypertelorism, downslanting palpebral fissures, blue sclerae, ear deformities, high palate) evident at birth or in early infancy, and characteristic cutaneous features like skin hyperextensibility, skin fragility with atrophic scars, easy bruising, and increased palmar wrinkling. Additional features include recurrent/chronic dislocations, chest and spinal deformities, peculiarly shaped fingers, colonic diverticula, pneumothorax, and urogenital and ophthalmological abnormalities, among others. Molecular testing is obligatory to confirm the diagnosis.
ORPHACODE:	2953
SYNONYMS:	Adducted thumb-clubfoot syndrome Distal arthrogryposis with peculiar facies and hydronephrosis Dündar syndrome Ehlers-Danlos syndrome, Kosho type Musculocontractural EDS mcEDS

XREF(S):	Orphanet ICD-10 OMIM OMIM
ANALYTE(S):	CHST14 DSE
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Ehlers-Danlos syndroom, EDS (gene panel)
- Epidermolysis bullosa (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Gent
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- carbohydrate sulfotransferase 14
- dermatan sulfate epimerase

Related Gene Panels

- Ehlers-Danlos syndrome -UGent
- Epidermolysis bullosa and bladder diseases (60 genes) - KUL
- Recessive Ehlers-Danlos Syndrome (11 genes) - UGent