GENETIC TEST:
Ehlers-Danlos syndroom, EDS (gene panel)

FULL NAME:	Ehlers-Danlos syndroom, EDS (gene panel)
DESCRIPTION:	ADAMTS2, AEBP1, B3GALT6, B3GAT3, B4GALT7, C1R, C1S, CHST14, COL12A1, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, DSE, FKBP14, PLOD1, PRDM5, RIN2, SLC39A13, XYLT1, XYLT2, ZNF469
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Mutation confirmation, Post-natal Diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA, DNA
METHOD CATEGORY:	Sequence analysis: entire coding region Deletion/duplication analysis
METHOD TECHNIQUE:	Next Generation Sequencing (NGS)
RIZIV CODE:	565493-565504
ACCREDITATION (ISO 15189):	2021-09-11 / 2026-09-10

TURNAROUND TIME (MAXIMUM):	4 months
CREATED:	11 Dec 2019 - 13:10
CHANGED:	31 Jan 2023 - 13:58
URL:	https://www.cmgg.be/nl/zorgverlener/labguide/constitutioneel-genetische-aandoen…

RELATED CONTENT

Related Diseases

- Arthrochalasia Ehlers-Danlos syndrome
- B3GALT6-related spondylodysplastic Ehlers-Danlos syndrome
- B4GALT7-related spondylodysplastic Ehlers-Danlos syndrome
- Bethlem myopathy
- Brittle cornea syndrome
- Cardiac-valvular Ehlers-Danlos syndrome
- Classical Ehlers-Danlos syndrome
- Classical-like Ehlers-Danlos syndrome type 2
- Congenital muscular dystrophy, Ullrich type
- Dermatosparaxis Ehlers-Danlos syndrome
- Desbuquois syndrome
- Ehlers-Danlos/osteogenesis imperfecta syndrome
- High bone mass osteogenesis imperfecta
- Kyphoscoliotic Ehlers-Danlos syndrome due to FKBP22 deficiency
- Kyphoscoliotic Ehlers-Danlos syndrome due to lysyl hydroxylase 1 deficiency
- Larsen-like syndrome, B3GAT3 type
- Musculocontractural Ehlers-Danlos syndrome
- Myopathic Ehlers-Danlos syndrome
- Periodontal Ehlers-Danlos syndrome
- RIN2 syndrome
- Reunion Island Larsen-like syndrome
- SLC39A13-related spondylodysplastic Ehlers-Danlos syndrome
- Vascular Ehlers-Danlos syndrome
- XYLT1-CDG

Related Laboratories

- Centrum Medische Genetica - UZ Gent

Related Analytes

- ADAM metallopeptidase with thrombospondin type 1 motif 2
- AE binding protein 1
- beta-1,3-galactosyltransferase 6
- beta-1,3-glucuronyltransferase 3
- beta-1,4-galactosyltransferase 7
- complement C1r
- complement C1s
- carbohydrate sulfotransferase 14
- collagen type XII alpha 1 chain
- collagen type I alpha 1 chain
- collagen type I alpha 2 chain
- collagen type III alpha 1 chain
- collagen type V alpha 1 chain
- collagen type V alpha 2 chain
- dermatan sulfate epimerase
- FKBP prolyl isomerase 14
- filamin A
- filamin B
- procollagen-lysine,2-oxoglutarate 5-dioxygenase 1
- PR/SET domain 5
- Ras and Rab interactor 2
- solute carrier family 39 member 13
- TGF-beta activated kinase 1 (MAP3K7) binding protein 2
- xylosyltransferase 1

- xylosyltransferase 2
- zinc finger protein 469

Related Gene Panels

- Ehlers-Danlos syndrome -UGent
