
DISEASE:
Cardiac-valvular Ehlers-Danlos syndrome

NAME:	Cardiac-valvular Ehlers-Danlos syndrome
DESCRIPTION:	A rare form of Ehlers-Danlos syndrome (EDS) characterized by soft skin, skin hyperextensibility, easy bruisability, atrophic scar formation, joint hypermobility and severe, progressive cardiac valvular defects comprising mitral and/or aortic valve insufficiency.
ORPHACODE:	230851
SYNONYMS:	Cardiac-valvular EDS cvEDS
XREF(S):	<u>Orphanet</u> <u>ICD-10</u> <u>OMIM</u>
ANALYTE(S):	<u>COL1A2</u>
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: <http://gentest.healthdata.be/disease/1583>

RELATED CONTENT

Related Genetic Tests

- [Aneurysm, Thoracic Aortic, familial \(gene panel\)](#)
- [Ehlers-Danlos syndroom, EDS \(gene panel\)](#)

Related Laboratories

- [Centrum Medische Genetica - UZ Antwerpen](#)
- [Centrum Medische Genetica - UZ Gent](#)

Related Analytes

- [collagen type I alpha 2 chain](#)

Related Gene Panels

- [Ehlers-Danlos syndrome -UGent](#)
- [Familial Thoracic Aortic Aneurysm \(genepanel\) - UZA](#)

Source URL: <http://gentest.healthdata.be/disease/1583>