sciensano **Belgian Genetic Tests database**

DISEASE:
Ebstein malformation of the tricuspid valve

NAME:	Ebstein malformation of the tricuspid valve
DESCRIPTION:	A rare congenital cardiac anomaly characterized by downward (apical) displacement of the functional annulus, due to incomplete delamination of the septal and inferior leaflets of the tricuspid valve such that they are hinged within the right ventricle, rather than as expected at the atrioventricular junction. The anterosuperior leaflet is often abnormal (redundancy, fenestrations, tethering with abnormal subvalvar apparatus). The atrioventricular junction and the "atrialized" portion of the right ventricle are dilated, with variable degrees of thinning of the right ventricular wall.
ORPHACODE:	1880
SYNONYMS:	Ebstein anomaly of the tricuspid valve
XREF(S):	Orphanet ICD-10 MedDRA OMIM
ANALYTE(S):	MYH7
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/318

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