DISEASE:
Coloboma of choroid and retina

NAME:	Coloboma of choroid and retina
DESCRIPTION:	Coloboma of choroid and retina is a rare, genetic developmental defect during embryogenesis characterized by the partial absence of retinal pigment epithelium and choroid, most frequently located in the inferonasal quadrant. Patients usually present reduced vision and have an increased risk for retinal detachment. Other ocular anomalies (e.g. coloboma of iris, microcornea, nystagmus, strabismus, microphthalmos) are usually associated, however it may also be isolated.
ORPHACODE:	98942
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	ACTG1 FZD5 SALL2 ABCB6 PAX6
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/3603

RELATED CONTENT

Related Genetic Tests

- Cataract (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Gent

Related Analytes

- ATP binding cassette subfamily B member 6 (Langereis blood group)
- actin gamma 1
- frizzled class receptor 5
- paired box 6
- spalt like transcription factor 2

Related Gene Panels

- Cataract - UGent
- test