DISEASE:
Cone rod dystrophy

NAME:	Cone rod dystrophy
DESCRIPTION:	A rare genetic isolated inherited retinal disorder characterized by primary cone degeneration with significant secondary rod involvement, with a variable fundus appearance. Typical presentation includes decreased visual acuity, central scotoma, photophobia, color vision alteration, followed by night blindness and loss of peripheral visual field.
ORPHACODE:	1872

XREF(S):	Orphanet OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM ICD-10 OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM OMIM

ANALYTE(S):	RPGR PRPH2 CFAP410 PITPNM3 OPN1MW CRX NMNAT1 CFAP418 CDHR1 ABCA4 RIMS1 RPGRIP1 CACNA1F CNGA3 AIPL1 GUCA1A GUCY2D OPN1LW RAX2 SEMA4A PROM1 CACNA2D4 ADAM9 UNC119 RAB28 POC1B DRAM2 TTLL5 TLCD3B ATF6
CREATED:	13 May 2019 - 01:02

CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/770

RELATED CONTENT

Related Genetic Tests

- Achromatopsia
- Ciliopathy (gene panel)
- Leber Congenital Amaurosis - Retinal dystrophy, early onset (gene panel)
- Retinitis pigmentosa, X-Linked

Related Laboratories

- Centrum Medische Genetica - UZ Gent

Related Analytes

- ATP binding cassette subfamily A member 4
- ADAM metallopeptidase domain 9
- aryl hydrocarbon receptor interacting protein like 1
- activating transcription factor 6
- calcium voltage-gated channel subunit alpha1 F
- calcium voltage-gated channel auxiliary subunit alpha2delta 4
- cadherin related family member 1
- cilia and flagella associated protein 410
- cilia and flagella associated protein 418
- cyclic nucleotide gated channel subunit alpha 3
- cone-rod homeobox
- DNA damage regulated autophagy modulator 2

- guanylate cyclase activator 1A
- guanylate cyclase 2D, retinal
- nicotinamide nucleotide adenylyltransferase 1
- opsin 1, long wave sensitive
- opsin 1, medium wave sensitive
- PITPNM family member 3
- POC1 centriolar protein B
- prominin 1
- peripherin 2
- RAB28, member RAS oncogene family
- retina and anterior neural fold homeobox 2
- regulating synaptic membrane exocytosis 1
- retinitis pigmentosa GTPase regulator
- RPGR interacting protein 1
- semaphorin 4A
- TLC domain containing 3B
- tubulin tyrosine ligase like 5
- unc-119 lipid binding chaperone

Related Gene Panels

- Achromatopsia (2 genes) - UGent
- Autosomal dominant Retinitis pigmentosa (11 genes) - UGent
- Ciliopathy (120 genes) - UGent
- Leber - Retinal, early onset
- Leber Congenital Amaurosis - UGent
- Retinitis pigmentosa, X-linked - UGent
