DISEASE:
Achromatopsia

NAME:	Achromatopsia
DESCRIPTION:	A rare autosomal recessive retinal disorder characterized by color blindness, nystagmus, photophobia, and severely reduced visual acuity due to the absence or impairment of cone function.
ORPHACODE:	49382
SYNONYMS:	ACHM Complete or incomplete color blindness Pingelapese blindness Rod monochromacy Rod monochromatism Total color blindness

XREF(S):	Orphanet MedDRA ICD-10 OMIM OMIM OMIM OMIM OMIM OMIM
ANALYTE(S):	PDE6C RPGR CNGA3 CNGB3 PDE6H GNAT2 ATF6
CREATED:	13 May 2019 - 01:02
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Source URL: http://gentest.healthdata.be/disease/3008

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- Ciliopathy (gene panel)

Related Laboratories

- Centrum Medische Genetica - UZ Gent

Related Analytes

- activating transcription factor 6
- cyclic nucleotide gated channel subunit alpha 3
- cyclic nucleotide gated channel subunit beta 3
- G protein subunit alpha transducin 2
- phosphodiesterase 6C
- phosphodiesterase 6H
- retinitis pigmentosa GTPase regulator

Related Gene Panels

- Achromatopsia (2 genes) - UGent
- Ciliopathy (120 genes) - UGent

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