

Table 1. Eye disease involved genes for which molecular diagnostic screening currently (29-01-2009) available.

(update: 29 january 2009)

1. Disease Name / Abbreviation	2. Locus	3. OMIM #	% tests, mutation identified in the gene
Amaurosis Congenita Leber's /LCA	CRB1	604210	about 13%
Amaurosis Congenita Leber's /LCA	RPE65	180069	about 10%
Amaurosis Congenita Leber's /LCA	CRX	602225	about 3%
Amaurosis Congenita Leber's /LCA	GUCY2D	600179	between 6-20%
Amaurosis Congenita Leber's /LCA	AIPL1	604392	about 6%
Amaurosis Congenita Leber's /LCA	RPGRIP1	605446	about 6%
Autosomal Dominant Pattern Dystrophy	RDS	179605	about 50%
Autosomal Dominant Retinitis Pigmentosa	RDS	179605	about 5%
Autosomal Dominant Retinitis Pigmentosa 4 / Rhodopsin	RHO	180380	about 10%
Autosomal recessive cone rod dystrophy 3/	ABCR	601691	about 60%
Autosomal recessive Retinitis Pigmentosa - 20	RPE65	180069	about 10%
Autosomal recessive Retinitis Pigmentosa -12	CRB1	600105	about 10%
Autosomal recessive Retinitis Pigmentosa -19	ABCR	601691	about 5%
Basal Laminar Drusen	CFH	126700	?
Dominant Optic atrophy	OPA1	165500	about 40%
Congenital Stationary nightblindness (CSNB1, complete form)	NYX	310500	?
Congenital Stationary nightblindness CSNB2, incomplete form)	CACNA1F	300071	?
Doyme Honeycomb retinal dystrophy	EFEMP1	126600	?
Best Macular dystrophy (early onset)	VMD2	153700	about 50%
Primary Open Angle Glaucoma	MYOC	601652	about 4%
Primary Open Angle Glaucoma	OPTN	602432	about 15%
Primary Congenital Glaucoma	CYP1B1	601771	
Pseudoxanthoma elasticum /PXE	ABCC6	603234	about 85%
Stargardt Disease	ABCR	601691	about 80%
Stargardt Disease 3 (AD)	ELOVL4	600110	?
X-linked cone dystrophy	RPGR ORF15	312610	about 50%
Oculocutaneous Albinism Type 1	OCA1, Tyr	606933	?
Oculocutaneous Albinism Type 2	OCA2, (P)	203200	?
X-linked ocular albinism	OA1	300500	about 60%
X-linked Retinitis Pigmentosa*	RPGR ORF15	312610	about 70%

X-linked Retinitis Pigmentosa *	RP2-linkage	312600	
X-linked Retinitis Pigmentosa *	RP3-linkage	312610	
X-linked Retinoschisis	XLRS1	312700	about 95%

* For X-linked Retinitis pigmentosa screening, we combine sequencing and linkage analysis. Please contact us prior of sending the samples to our laboratory about the requirements.

1 Report time of the molecular diagnostic tests will usually be about 6-12 months. Screening strategy in general direct sequencing of the entire coding region of the gene requested.

2 For additional information, please contact:

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3 All molecular diagnoses are evaluated under the supervision and final responsibility of registered clinical molecular geneticists (Prof. Dr. A. Bergen and Dr M. Mannens).

4 All molecular diagnoses are performed using the guidelines as determined by the workgroup of the [Landelijk Overleg DNA diagnostiek \(LOD\)](#). Accreditation of the laboratory is being obtained in cooperation with the AMC.

5 Nationally, the investigation is funded by the patient's health insurance. Alternatively, independent clinics and private patients or requests from countries other than the Netherlands may pay directly.

6 Thorough clinical investigation of patients by a certified ophthalmologist, preferably an ophthalmogeneticist is essential. Intake, evaluation and interpretation of the molecular genetic results should be presented to the patient/family only by a qualified genetic counsellor so that unnecessary confusion and stress might be avoided. Also presymptomatic investigation to determine the genetic status of an individual (*e.g.*, carrier status), including prenatal diagnostics, requires prior genetic counselling.

7 Direct patient consultation is not provided.

Further information about cloned and/or mapped genes causing retinal diseases can be found at <http://www.sph.uth.tmc.edu/Retnet/disease.htm> . Also information for

several disease genes is presented on the genereviews at the Genetests website:
<http://www.genetests.org/> .