Medizinische Genetik Mainz

Genetic analyses for hereditary eye diseases

Hereditary eye diseases are characterised by strong genetic heterogeneity and overlapping clinical presentations. Molecular genetic testing often helps establish a specific diagnosis and, in the context of clinical assessment, predict the expected progression of the disorder and initiate targeted treatment.

Retinal diseases

A	Achromatopsia
C	Cone-rod dystrophy
F	Flecked retina syndrome
L	Leber congenital amaurosis (LCA)
	Leber hereditary optic neuropathy (LHON)
N	Night blindness, congenital stationary X-linked
	Nystagmus
O	Optic atrophy
P	Peroxisome biogenesis disorder
R	Retinitis pigmentosa
	Retinoblastoma
S	Stargardt disease
U	Usher syndrome type 1
	Usher syndrome type 2

(Peripheral) eye diseases

A	Albinism
	Anterior segment dysgenesis (ASD) disorders
C	Cataract
	Chediak-Higashi syndrome
	Congenital fibrosis of the external eye muscles (CFEOM)
	Corneal dystrophy
F	Familial exudative vitreoretinopathy
G	Glaucoma
	Griscelli syndrome
H	Hermansky-Pudlak syndrome
M	Microphthalmia
R	Retinal detachment

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W	WAGR syndrome (Wilms tumor aniridia syndrome)

Syndromic diseases with eye involvement

A	Alagille syndrome
	Alport syndrome
B	Bardet-Biedl syndrome
C	CHARGE syndrome
H	Hyperoxaluria
J	Joubert syndrome
M	Mainzer-Saldino syndrome
N	Norrie syndrome
R	Refsum disease
S	Senior-Loken syndrome
	Stickler syndrome
T	Tuberous Sclerosis Complex
W	Waardenburg syndrome