Sub-thematic areas of expertise included in the main thematic group of rare or complex disease(s) or condition(s) covered by ERN-EYE with Specific Code/ICD/Orphanet classification(s), if available.

Sub-thematic Areas of Expertise	Rare or Complex Disease(s), Condition(s) or Highly Specialised Intervention(s)	Code / ICD / Orphacode Group of codes*	Incidence (Number of cases/year)	Prevalence **
Retinal Dystrophy	Retinis Pigmentosa	ORPHA791	Around 5000 per year (estimation)	1/3 000 – 1/5 000
Retinal Dystrophy	Leber congenital amaurosis	ORPHA65		1/50 000 - 1/33 000
Retinal Dystrophy	Syndromic retinitis pigmentosa	ORPHA98661		Group of diseases, prevalence unavailable
Retinal Dystrophy	Metabolic disease associated with ocular features	ORPHA98710		Group of diseases, prevalence unavailable
Retinal Dystrophy	Metabolic disease with pigmentary retinitis	ORPHA98713		Group of diseases, prevalence unavailable
Retinal Dystrophy	Cohen syndrome	ORPHA193		Unknown
Retinal Dystrophy	Joubert syndrome with oculorenal defect	ORPHA2318		Unknown
Retinal Dystrophy	Usher syndrome	ORPHA886		1/30 000
Retinal Dystrophy	Alström syndrome	ORPHA64		Unknown
Retinal Dystrophy	Bardet-Biedl syndrome	ORPHA110		1/125 000 - 1/175 000
Retinal Dystrophy	Choroideremia	ORPHA180		1/50 000 - 1/100 000
Retinal Dystrophy	Genetic macular dystrophy	ORPHA98664		Group of diseases, prevalence unavailable
Retinal Dystrophy	Progressive cone dystrophy	ORPHA1871		Unknown
Retinal Dystrophy	Best vitelliform macular dystrophy	ORPHA1243		1/5 000 - 1/67 000
Retinal Dystrophy	Patterned dystrophy of the retinal pigment epithelium	ORPHA63454		Group of diseases, prevalence unavailable
Retinal Dystrophy	Colobomatous and areolar dystrophy	ORPHA98665		Group of diseases, prevalence unavailable
Retinal Dystrophy	Cone rod dystrophy	ORPHA1872		1/40 000
Retinal Dystrophy	Familial flecked retinopathy	ORPHA227786		Group of diseases, prevalence unavailable
Retinal Dystrophy	Stargardt disease	ORPHA827		1/8 000 - 1/10 000
Retinal Dystrophy	Color-vision disease	ORPHA98658		Group of diseases, prevalence unavailable
Retinal Dystrophy	Achromatopsia	ORPHA49382		1/30 000 - 1/50 000
Retinal Dystrophy	Genetic vitreous-retinal disease	ORPHA98657		Group of diseases, prevalence unavailable
Retinal Dystrophy	Stickler syndrome	ORPHA828		Unknown
Retinal Dystrophy	X-linked retinoschisis	ORPHA792		1/5 000- 1/25,000
Retinal Dystrophy	Familial exudative vitreoretinopathy	ORPHA891		Unknown
Retinal Dystrophy	Congenital stationary night blindness	ORPHA215		Unknown

Neuro-ophthalmology and rare eye diseases	Hereditary optic neuropathy	ORPHA98671	Around 650 per year (estimation)	Group of diseases, prevalence unavailable
Neuro-ophthalmology and rare eye diseases	Leber hereditary optic neuropathy	ORPHA104		1/15 000 - 1/50 000
Neuro-ophthalmology and rare eye diseases	Autosomal dominant optic atrophy	ORPHA98672		Group of diseases, prevalence unavailable
Neuro-ophthalmology and rare eye diseases	Autosomal recessive isolated optic atrophy	ORPHA98676		Unknown
Neuro-ophthalmology and rare eye diseases	Syndromic optic nerve hypoplasia	ORPHA137905		Group of diseases, prevalence unavailable
Neuro-ophthalmology and rare eye diseases	Syndromic hereditary optic neuropathy	ORPHA441434		Group of diseases, prevalence unavailable
Neuro-ophthalmology and rare eye diseases	Metabolic disease associated with ocular features	ORPHA98710		Group of diseases, prevalence unavailable
Neuro-ophthalmology and rare eye diseases	Mitochondrial disease with eye involvement	ORPHA98695		Group of diseases, prevalence unavailable
Neuro-ophthalmology and rare eye diseases	Leigh syndrome	ORPHA506		1/36 000
Neuro-ophthalmology and rare eye diseases	Neuro-ophthalmological disease	ORPHA140653		Group of diseases, prevalence unavailable
Neuro-ophthalmology and rare eye diseases	Craniostenosis associated with a strabismus	ORPHA98684		Group of diseases, prevalence unavailable
Neuro-ophthalmology and rare eye diseases	Oculomotor palsy	ORPHA98685		Group of diseases, prevalence unavailable
Neuro-ophthalmology and rare eye diseases	Supranuclear oculomotor palsy	ORPHA98687		Group of diseases, prevalence unavailable
Neuro-ophthalmology and rare eye diseases	Nuclear oculomotor paralysis	ORPHA100932		Group of diseases, prevalence unavailable
Neuro-ophthalmology and rare eye diseases	Oculomotor apraxia or related oculomotor disease	ORPHA98688		Group of diseases, prevalence unavailable
Neuro-ophthalmology and rare eye diseases	Myopathy with eye involvement	ORPHA98689		Group of diseases, prevalence unavailable
Neuro-ophthalmology and rare eye diseases	Abnormal eye movements	ORPHA98691		Group of diseases, prevalence unavailable
Neuro-ophthalmology and rare eye diseases	Nervous system anomaly with eye involvement	ORPHA98692		Group of diseases, prevalence unavailable
Neuro-	Rare acquired eye	ORPHA101949		Group of

ophthalmology and rare eye diseases	disease			diseases, prevalence unavailable
Neuro-ophthalmology and rare eye diseases	Balhint syndrom	ORPHA363746		Unknown
Neuro-ophthalmology and rare eye diseases	Holmes-Adie syndrome	ORPHA454718		Unknown
Neuro-ophthalmology and rare eye diseases	Progressive supranuclear palsy syndrome	ORPHA683		About 1/16600
Neuro-ophthalmology and rare eye diseases	Atypical Progressive supranuclear palsy syndrome	ORPHA99750		Unknown
Neuro-ophthalmology and rare eye diseases	Classic Progressive supranuclear palsy syndrome	ORPHA240071		About 1/16600
Neuro-ophthalmology and rare eye diseases	Progressive supranuclear palsy-pure akinesia with gait freezing syndrome	ORPHA240094		Unknown
Neuro-ophthalmology and rare eye diseases	Progressive supranuclear palsy-progressive non-fluent aphasia syndrome	ORPHA240112		Unknown (<10 cases reported in the literature)
Neuro-ophthalmology and rare eye diseases	Progressive supranuclear palsy-parkinsonism syndrome	ORPHA240085		Unknown
Neuro-ophthalmology and rare eye diseases	Progressive supranuclear palsy-corticobasal syndrome	ORPHA240103		About 1/16600
Neuro-ophthalmology and rare eye diseases	Ptosis	ORPHA98578		Group of diseases, prevalence unavailable
Neuro-ophthalmology and rare eye diseases	Carnevale syndrome (	ORPHA2998		Unknown
Neuro-ophthalmology and rare eye diseases	Marcus-Gunn syndrome	ORPHA91412		Unknown
Neuro-ophthalmology and rare eye diseases	Septopreoptic holoprosencephaly	ORPHA280195		Unknown
Neuro-ophthalmology and rare eye diseases	Spastic paraplegia-optic atrophy-neuropathy syndrome	ORPHA320406		2 to 6 /100000
Neuro-ophthalmology and	Syndromic hereditary optic neuropathy	ORPHA441434		Group of diseases, prevalence

rare eye diseases				unavailable
Neuro-ophthalmology and rare eye diseases	Syndromic optic nerve hypoplasia	ORPHA137905		Group of diseases, prevalence unavailable
Neuro-ophthalmology and rare eye diseases	Treft-Sanborn-Carey syndrome	ORPHA3349		Unknown
Neuro-ophthalmology and rare eye diseases	Neuromyelitis optica	ORPHA71211		1 to 2 / 100000
Pediatric Ophthalmology Rare Diseases	Hereditary glaucoma	ORPHA359	Around 1800 patients per year (estimation)	Unknown
Pediatric Ophthalmology Rare Diseases	Iridogoniodysgenesis	ORPHA98634		Group of diseases, prevalence unavailable
Pediatric Ophthalmology Rare Diseases	Axenfeld-Rieger syndrome	ORPHA782		1/200 000
Pediatric Ophthalmology Rare Diseases	Aniridia	ORPHA77		1/64 000 - 1/96 000
Pediatric Ophthalmology Rare Diseases	Syndromic aniridia	ORPHA98557		Group of diseases, prevalence unavailable
Pediatric Ophthalmology Rare Diseases	Isolated aniridia	ORPHA250923		1/64 000 - 1/96 000
Pediatric Ophthalmology Rare Diseases	Peters anomaly	ORPHA708		<1/1 000 000
Pediatric Ophthalmology Rare Diseases	Congenital glaucoma	ORPHA98976		1/27 800
Pediatric Ophthalmology Rare Diseases	Juvenile glaucoma	ORPHA98977		1/50 000
Pediatric Ophthalmology Rare Diseases	Developmental defect of the eye	ORPHA98553		Group of diseases, prevalence unavailable
Pediatric Ophthalmology Rare Diseases	Anophthalmia-microphthalmia syndrome	ORPHA98555		Group of diseases, prevalence unavailable
Pediatric Ophthalmology Rare Diseases	Syndromic microphthalmia	ORPHA202948		Group of diseases, prevalence unavailable
Pediatric Ophthalmology Rare Diseases	Ocular coloboma	ORPHA194		Group of diseases, prevalence unavailable
Pediatric Ophthalmology Rare Diseases	Septo-optic dysplasia spectrum	ORPHA3157		Unknown
Pediatric Ophthalmology Rare Diseases	Rare palpebral, lacrimal system and conjunctival disease	ORPHA98559		Group of diseases, prevalence

				unavailable
Pediatric Ophthalmology Rare Diseases	Rare palpebral disease	ORPHA98560		Group of diseases, prevalence unavailable
Pediatric Ophthalmology Rare Diseases	Congenital Ptosis	ORPHA91411		Unknown
Pediatric Ophthalmology Rare Diseases	Rare lacrimal system disease	ORPHA98602		Group of diseases, prevalence unavailable
Pediatric Ophthalmology Rare Diseases	Lens and zonula anomaly	ORPHA98639		Group of diseases, prevalence unavailable
Pediatric Ophthalmology Rare Diseases	Early-onset non-syndromic cataract	ORPHA91492		Unknown
Pediatric Ophthalmology Rare Diseases	Syndromic cataract	ORPHA98641		Group of diseases, prevalence unavailable
Pediatric Ophthalmology Rare Diseases	Systemic disease with cataract	ORPHA98643		Group of diseases, prevalence unavailable
Pediatric Ophthalmology Rare Diseases	Genetic vitreous-retinal disease	ORPHA98657		Group of diseases, prevalence unavailable
Pediatric Ophthalmology Rare Diseases	Norrie disease	ORPHA649		Unknown
Pediatric Ophthalmology Rare Diseases	Coats plus syndrome	ORPHA313838		<1/1 000 000
Pediatric Ophthalmology Rare Diseases	Vitreoretinal degeneration	ORPHA98670		Group of diseases, prevalence unavailable
Pediatric Ophthalmology Rare Diseases	X-linked retinoschisis	ORPHA792		1/5 000 - 1/25 000
Pediatric Ophthalmology Rare Diseases	Familial exudative vitreoretinopathy	ORPHA891		Unknown
Pediatric Ophthalmology Rare Diseases	Genodermatosis with ocular features	ORPHA98696		Group of diseases, prevalence unavailable
Pediatric Ophthalmology Rare Diseases	Pigmentation disorder with eye involvement	ORPHA98700		Group of diseases, prevalence unavailable
Pediatric Ophthalmology Rare Diseases	Oculocutaneous albinism	ORPHA55		1/17 000
Pediatric Ophthalmology Rare Diseases	Ocular albinism	ORPHA284804		Group of diseases, prevalence

				unavailable
Pediatric Ophthalmology Rare Diseases	Syndromic oculocutaneous albinism	ORPHA284811		Group of diseases, prevalence unavailable
Pediatric Ophthalmology Rare Diseases	Pigmentation disorder with eye involvement excluding Albinism	ORPHA98708		Group of diseases, prevalence unavailable
Pediatric Ophthalmology Rare Diseases	Phakomatosis with eye involvement	ORPHA98701		Group of diseases, prevalence unavailable
Pediatric Ophthalmology Rare Diseases	Connective tissue disease with eye involvement	ORPHA98702		Group of diseases, prevalence unavailable
Pediatric Ophthalmology Rare Diseases	Marfan syndrome	ORPHA558		1/5 000
Pediatric Ophthalmology Rare Diseases	Metabolic disease associated with ocular features	ORPHA98710		Group of diseases, prevalence unavailable
Pediatric Ophthalmology Rare Diseases	Mitochondrial disease with eye involvement	ORPHA98695		Group of diseases, prevalence unavailable
Anterior Segment Rare Eye Diseases	Hereditary glaucoma	ORPHA359	Around 1100 patients per year (estimation)	Unknown
Anterior Segment Rare Eye Diseases	Juvenile glaucoma	ORPHA98977		1/50 000
Anterior Segment Rare Eye Diseases	Corneal dystrophy	ORPHA34533		Unknown
Anterior Segment Rare Eye Diseases	Superficial corneal dystrophy	ORPHA98625		Unknown
Anterior Segment Rare Eye Diseases	Stromal corneal dystrophy	ORPHA98626		Unknown
Anterior Segment Rare Eye Diseases	Posterior corneal dystrophy	ORPHA98627		Unknown
Anterior Segment Rare Eye Diseases	Syndromic corneal dystrophy	ORPHA98628		Group of diseases, prevalence unavailable
Anterior Segment Rare Eye Diseases	Rare conjunctival disease	ORPHA98610		Group of diseases, prevalence unavailable
Anterior Segment Rare Eye Diseases	Rare refraction anomaly	ORPHA98618		Group of diseases, prevalence unavailable
Anterior Segment Rare Eye Diseases	Rare disease with glaucoma as a major feature	ORPHA98638		Group of diseases, prevalence unavailable
Anterior Segment Rare Eye Diseases	Lens and zonula anomaly	ORPHA98639		Group of diseases, prevalence

				unavailable
Anterior Segment Rare Eye Diseases	Connective tissue disease with eye involvement	ORPHA98702		Group of diseases, prevalence unavailable
Anterior Segment Rare Eye Diseases	Marfan syndrome	ORPHA558		1/5 000
Anterior Segment Rare Eye Diseases	Metabolic disease with corneal opacity	ORPHA98711		Group of diseases, prevalence unavailable
Anterior Segment Rare Eye Diseases	Metabolic disease with cataract	ORPHA98712		Group of diseases, prevalence unavailable
Anterior Segment Rare Eye Diseases	Keratoconus	ORPHA156071		Group of diseases, prevalence unavailable
Anterior Segment Rare Eye Diseases	Syndromic keratoconus	ORPHA98623		Group of diseases, prevalence unavailable
Anterior Segment Rare Eye Diseases	Rare acquired eye disease	ORPHA101949		Group of diseases, prevalence unavailable