



## **ERN-EYE**

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	Rare or Complex	Code / ICD /	Incidence	Prevalence
of Expertise	Disease(s), Condition(s)	Orphacode	(Number of	**
	or Highly Specialised	Group of	cases/year)	
	Intervention(s)	codes*		
Retinal Dystrophy	Retinis Pigmentosa	ORPHA791	Around 5000	1/3 000 -
Datinal Dustranbu	Lobor congonital	ORPHA65	per year (estimation)	1/5 000 1/50 000 -
Retinal Dystrophy	Leber congenital amaurosis		(estillation)	1/33 000
Retinal Dystrophy	Syndromic retinitis	ORPHA98661		Group of diseases,
	pigmentosa			prevalence
				unavailable
Retinal Dystrophy	Metabolic disease	ORPHA98710		Group of
	associated with ocular			diseases, prevalence
	features			unavailable
Retinal Dystrophy	Metabolic disease with	ORPHA98713	1	Group of
	pigmentary retinitis			diseases,
				prevalence unavailable
Retinal Dystrophy	Cohen syndrome	ORPHA193		Unknown
Retinal Dystrophy	Joubert syndrome with	ORPHA2318		Unknown
riceman 2 year opiny	oculorenal defect	0		
Retinal Dystrophy	Usher syndrome	ORPHA886	1	1/30 000
Retinal Dystrophy	Alström syndrome	ORPHA64		Unknown
Retinal Dystrophy	Bardet-Biedl syndrome	ORPHA110		1/125 000 -
				1/ <mark>17</mark> 5 000
Retinal Dystrophy	Choroideremia	ORPHA180		1/50 000 -
				1/100 000
Retinal Dystrophy	Genetic macular	ORPHA98664		Group of
	dystrophy			diseases, prevalence
				unavailable
Retinal Dystrophy	Progressive cone	ORPHA1871		Unknown
	dystrophy			
Retinal Dystrophy	Best vitelliform macular	ORPHA1243		1/5 000 -
	dystrophy			1/67 000
Retinal Dystrophy	Patterned dystrophy of	ORPHA63454		Group of diseases,
	the retinal pigment			prevalence
	epithelium			unavailable
Retinal Dystrophy	Colobomatous and	ORPHA98665		Group of
	areolar dystrophy			diseases, prevalence
				unavailable
Retinal Dystrophy	Cone rod dystrophy	ORPHA1872	1	1/40 000
Retinal Dystrophy	Familial flecked	ORPHA227786	1	Group of
, , ,	retinopathy			diseases,
				prevalence
Retinal Dystrophy	Stargardt disease	ORPHA827	1	unavailable 1/8 000 -
Retinal Dystrophy	Stargarut disease	ONI HAOZ7		1/10 000
Retinal Dystrophy	Color-vision disease	ORPHA98658		Group of
riceman 2 your opiny	delet tistert discuse	<b>3</b>		diseases,
				prevalence
Datinal Dustranhu	Ashramatansia	ODDUA 40202	-	unavailable
Retinal Dystrophy	Achromatopsia	ORPHA49382		1/30 000 - 1/50 000
Retinal Dystrophy	Genetic vitreous-retinal	ORPHA98657		Group of
Retinal Dystrophly	disease	JN 11A30037		diseases,
	alocase			prevalence
				unavailable
Retinal Dystrophy	Stickler syndrome	ORPHA828		Unknown
Retinal Dystrophy	X-linked retinoschisis	ORPHA792		1/5 000-
Dational Date 1	Familialday	ODDUAGOS		1/25,000
Retinal Dystrophy	Familial exudative	ORPHA891		Unknown
Retinal Dystrophy	vitreoretinopathy Congenital stationary	ORPHA215	1	Unknown
recinal bystrophry	night blindness	OW 110513		CHRIDWII
	THETTE DIFFICULTEDS	1	1	1

Neuro- ophthalmology and	Hereditary optic neuropathy	ORPHA98671	Around 650 per year	Group of diseases, prevalence	
Neuro- ophthalmology and	Leber hereditary optic neuropathy	ORPHA104	(estimation)	unavailable 1/15 000 - 1/50 000	
rare eye diseases  Neuro-	Autosomal dominant	ORPHA98672	<u> </u>	Group of	
ophthalmology and rare eye diseases	optic atrophy			diseases, prevalence unavailable	
Neuro- ophthalmology and	Autosomal recessive isolated optic atrophy	ORPHA98676	-	Unknown	<b>×</b>
nare eye diseases  Neuro- ophthalmology and	Syndromic optic nerve hypoplasia	ORPHA137905		Group of diseases,	
rare eye 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	
Neuro- ophthalmology and rare eye diseases	Mitochondrial disease with eye involvement	ORPHA98695		unavailable 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	7(	Group of diseases, prevalence unavailable	
Neuro- ophthalmology and rare eye diseases	Craniostenosis associated with a strabismus	ORPHA98684	0	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	

	Ι		1	1.	1
ophthalmology and	disease			diseases,	
rare eye diseases				prevalence unavailable	
				diavallable	
Neuro-	Balhint syndrom	ORPHA363746		Unknown	
ophthalmology and					
rare eye diseases					
·					
Neuro-	Holmes-Adie syndrome	ORPHA454718		Unknown	1
ophthalmology and					<b>A</b>
rare eye diseases					
Neuro-	Progressive	ORPHA683		About	
ophthalmology and	supranuclear palsy syndrome			1/16600	
rare eye diseases	Syndrome				
Name	At a last December	00011400750	_	Unknown	
Neuro-	Atypical Progressive supranuclear palsy	ORPHA99750		Unknown	
ophthalmology and	syndrome				
rare eye diseases					
Neuro-	Classic Progressive	ORPHA240071	_	About	-
ophthalmology and	supranuclear palsy	ON 11A240071		1/16600	
rare eye diseases	syndrome				
. s. c cyc diocuses					
Neuro-	Progressive	ORPHA240094		Unknown	
ophthalmology and	supranuclear palsy-pure				
rare eye diseases	akinesia with gait				
•	freezing syndrome				
Neuro-	Progressive	ORPHA240112		Un <mark>kn</mark> own	1
ophthalmology and	supranuclear palsy-			(<10 cases	
rare eye diseases	progressive non-fluent			reported in the	
	aphasia syndrome			literature)	
				,	
Neuro-	Progressive	ORPHA240085		Unknown	1
ophthalmology and	supranuclear palsy-				
rare eye diseases	parkinsonism syndrome				
Neuro-	Progressive	ORPHA240103		About	
ophthalmology and	supranuclear palsy- corticobasal syndrome			1/16600	
rare eye diseases	corticobasar syndrome	·			
Name	Dhasia	00011400570		Group of	
Neuro- ophthalmology and	Ptosis	ORPHA98578		diseases,	
rare eye diseases				prevalence	
rare eye diseases				unavailable	
Neuro-	Carnevale syndrome (	ORPHA2998		Unknown	
ophthalmology and					
rare eye diseases					
Name	Marana Coma and dama	ODDIIA01413	_	Unknown	-
Neuro-	Marcus-Gunn syndrome	ORPHA91412		Unknown	
ophthalmology and rare eye diseases					
rare eye diseases					
Neuro-	Septopreoptic	ORPHA280195	-	Unknown	1
ophthalmology and	holoprosencephaly	0111111200133			
rare eye diseases					
,					
Neuro-	Spastic paraplegia-optic	ORPHA320406		2 to 6	1
ophthalmology and	atrophy-neuropathy			/100000	
rare eye diseases	syndrome				
Neuro-	Syndromic hereditary	ORPHA441434		Group of	
ophthalmology and	optic neuropathy			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	Unknown	
Pediatric Ophthalmology Rare Diseases	Iridogoniodysgenesis	ORPHA98634	(estimation)	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	1	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		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	1
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	70	Group of diseases, prevalence unavailable	
Pediatric Ophthalmology Rare Diseases	Genetic vitreous-retinal disease	ORPHA98657	O.	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	1/17 000	
Pediatric Ophthalmology Rare Diseases	Ocular albinism	ORPHA284804		Group of diseases, prevalence	

				unavailable	1
Pediatric	Syndromic	ORPHA284811		Group of	
Ophthalmology	oculocutaneous albinism			diseases,	
Rare Diseases				prevalence unavailable	
				unavanabic	
Pediatric	Pigmentation disorder	ORPHA98708	1	Group of	
Ophthalmology	with eye involvement			diseases,	
Rare Diseases	excluding Albinism			prevalence	
				unavailable	
Pediatric	Phakomatosis with eye	ORPHA98701		Group of	
Ophthalmology	involvement			diseases,	
Rare Diseases				prevalence	
				unavailable	
Pediatric	Connective tissue	ORPHA98702		Group of	
Ophthalmology	disease with eye	0		diseases,	
Rare Diseases	involvement			prevalence	
				unavailable	
Podiatris	Marfan sundrama	ORPHA558	4	1/5 000	
Pediatric Ophthalmology	Marfan syndrome	ONFIIADO		1/3 000	
Rare Diseases				_	
Pediatric	Metabolic disease	ORPHA98710	]	Group of	
Ophthalmology	associated with ocular			diseases,	
Rare Diseases	features			prevalence unavailable	
				unavanable	
Pediatric	Mitochondrial disease	ORPHA98695		Group of	
Ophthalmology	with eye involvement			diseases,	
Rare Diseases				prevalence	
			4	unavailable	
Anterior Segment	Hereditary glaucoma	ORPHA359	Around 1100	Unknown	
Rare Eye Diseases			patients per		
Anterior Segment Rare Eye Diseases	Juvenile glaucoma	ORPHA98977	year (estimation)	1/50 000	
Anterior Segment	Corneal dystrophy	ORPHA34533	(estimation)	Unknown	1
Rare Eye Diseases					
Anterior Segment	Superficial corneal	ORPHA98625		Unknown	
Rare Eye Diseases	dystrophy	ODDIMAGGGG	-	I I a lua a coma	-
Anterior Segment Rare Eye Diseases	Stromal corneal dystrophy	ORPHA98626		Unknown	
Anterior Segment	Posterior corneal	ORPHA98627	1	Unknown	1
Rare Eye Diseases	dystrophy				
Anterior Segment	Syndromic corneal	ORPHA98628		Group of	
Rare Eye Diseases	dystrophy			diseases, prevalence	
				unavailable	
Anterior Segment	Rare conjunctival	ORPHA98610	1	Group of	
Rare Eye Diseases	disease			diseases,	
				prevalence	
				unavailable	
Anterior Segment	Rare refraction anomaly	ORPHA98618	1	Group of	
Rare Eye Diseases				diseases,	
				prevalence	
				unavailable	
Anterior Segment	Rare disease with	ORPHA98638	-	Group of	-
Rare Eye Diseases	glaucoma as a major	UNFFIA98038		diseases,	
e Lye Discuses	feature			prevalence	
		1	1	unavailable	
			_		
Anterior Segment Rare Eye Diseases	Lens and zonula anomaly	ORPHA98639		Group of diseases,	

Anterior Segment Rare Eye Diseases			<u>_</u>		
Rare Eye Diseases	Connective tissue	ORPHA98702		Group of	
	disease with eye			diseases,	
	involvement			prevalence unavailable	
				unavallable	
Anterior Segment Rare Eye Diseases	Marfan syndrome	ORPHA558		1/5 000	
Anterior Segment	Metabolic disease with	ORPHA98711		Group of	
Rare Eye Diseases	corneal opacity			diseases,	
				prevalence	
				unavailable	
Anterior Segment	Metabolic disease with	ORPHA98712	<del> </del>	Group of	
Rare Eye Diseases	cataract			diseases,	
				prevalence	
				unavailable	
Anterior Segment	Keratoconus	ORPHA156071	<del> </del>	Group of	
Rare Eye Diseases				diseases,	
,				prevalence	
			l l	unavailable	
Anterior Segment	Syndromic keratoconus	ORPHA98623	1	Group of	
Rare Eye Diseases	,			diseases,	
,				prevalence	
				unavailable	
Anterior Segment	Rare acquired eye	ORPHA101949	-	Group of	
Rare Eye Diseases	disease	ON 11/4101545		diseases,	
naic Lyc Discuses	discuse			prevalence	
				unavailable	