

Disease

RPE65 associated Leber congenital amaurosis, early-onset severe retinal dystrophy¹

Gene

RPE65

Inheritance	AR
Current Treatments	gene therapy (Luxturna)
Evidence for Treatment	expert opinion
Non-Molecular Testing Availability	yes
Non-Molecular Test	full-field electroretinography and peripheral fundus autofluorescence ¹
ClinGen	https://actionability.clinicalgenome.org/ac/Pediatric/ui/stg2SummaryRpt?doc=AC012
Disease Frequency (per 100,000)	0.685 ²
Genes Included in Disease Frequency	RPE65
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=rpe65&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/books/NBK549574/>

² <https://pubmed.ncbi.nlm.nih.gov/35098484/>