

Disease

Hereditary retinoblastoma^{1, 2}

Gene

RB1

Inheritance	AD
Current Treatments	enucleation; cryotherapy; laser, systemic, or local ocular chemotherapy including intra-arterial chemotherapy, combined with or followed by laser or cryotherapy; radiation therapy using episcleral plaques; external beam radiotherapy
Evidence for Treatment	guideline ²
Non-Molecular Testing Availability	yes
Non-Molecular Test	ophthalmologic examination
ClinGen	https://actionability.clinicalgenome.org/ac/Pediatric/ui/stg2SummaryRpt?doc=AC001
Disease Frequency (per 100,000)	5.835 ¹
Genes Included in Disease Frequency	RB
Experimental / Investigational Therapies	https://clinicaltrials.gov/ct2/results?cond=Retinoblastoma&term=&cntry=&state=&city=&dist=

References

¹ <https://www.ncbi.nlm.nih.gov/books/NBK1452/>

² <https://pubmed.ncbi.nlm.nih.gov/26389442/>