RB1 gene

Associated Syndrome Name: Hereditary retinoblastoma

RB1 Summary Cancer Risk Table

CANCER	GENETIC CANCER RISK
Other	High Risk
Skin	Elevated Risk

RB1 gene Overview

Hereditary retinoblastoma [1, 2, 3, 4]

- Individuals with *RB1* mutations have hereditary retinoblastoma, a cancer that develops in the retina of the eye.
- Nearly all individuals with *RB1* mutations will develop retinoblastoma before the age of 5.
- There is a significantly increased risk for second primary cancers after retinoblastoma, most commonly sarcomas (cancers of the soft tissue or bone) or melanomas. The risk for these cancers appears to be higher in individuals who received high-dose radiation therapy to treat their retinoblastoma.
- There are rare mutations within the *RB1* gene that may be associated with lower risks for retinoblastoma and other cancers. **If this patient's *RB1* mutation is known to be associated with lower cancer risks, that information is provided in the \u201cDETAILS ABOUT: *RB1*\u201d section of the Genetic Result.**
- Although there are increased risks for cancer in individuals with *RB1* mutations, there are interventions that may reduce these risks. Guidelines from the American Academy of Ophthalmology (AAO) and American Association for Cancer Research (AACR) are listed below. It is recommended that individuals with *RB1* mutations be managed by a multidisciplinary team with expertise in medical genetics and the prevention and treatment of the cancers associated with this condition.

RB1 gene Cancer Risk Table

CANCER TYPE	AGE RANGE	CANCER RISK	RISK FOR GENERAL POPULATION
Retinoblastoma	To age 5[1, 2]	Up to 99%	<0.1%
Overall Cancer Risk	Risk for a second primary cancer within 40 years of retinoblastoma diagnosis[3, 5]	28%-41%	NA
Sarcoma	To age 80[1, 3, 5, 6, 7, 8]	High risk	0.4%
Melanoma	To age 80[1, 3, 5, 7, 8]	Elevated risk	1.6%

RB1 Cancer Risk Management Table

The overview of medical management options provided is a summary of professional society guidelines. The most recent version of each guideline should be consulted for more detailed and up-to-date information before developing a treatment plan for a particular patient.

This overview is provided for informational purposes only and does not constitute a recommendation. While the medical society guidelines summarized herein provide important and useful information, medical management decisions for any particular patient should be made in consultation between that patient and his or her healthcare provider and may differ from society guidelines based on a complete understanding of the patient's personal medical history, surgeries and other treatments.

CANCER TYPE	PROCEDURE	AGE TO BEGIN	FREQUENCY (UNLESS OTHERWISE INDICATED BY FINDINGS)
Overall Cancer Risk	Provide education about the signs and symptoms of cancer.[9]	From birth	Annually
	Physical examination[9]	From birth	Annually

CANCER TYPE	PROCEDURE	AGE TO BEGIN	FREQUENCY (UNLESS OTHERWISE INDICATED BY FINDINGS)
Retinoblastoma	Eye exam with dilation, preferably performed by an ophthalmologist with experience in retinoblastoma[2, 9]	From birth	Varies depending on age; refer to AAO guidelines.
	Consider brain MRI.[4]	At time of diagnosis of hereditary retinoblastoma	Every 6 months until age 4
Sarcoma	Consider whole-body MRI.[4, 9]	6 to 8 years, when general anesthesia is not required	Annually
Melanoma	Skin exam by pediatrician or dermatologist[9]	From birth	Annually

Information for Family Members

The following information for Family Members will appear as part of the MMT for a patient found to have a mutation in the RB1 gene.

This patient's relatives are at risk for carrying the same mutation(s) and associated cancer risks as this patient. Cancer risks for females and males who have this/these mutation(s) are provided below.

Family members should talk to a healthcare provider about genetic testing. Close relatives such as parents, children, brothers and sisters have the highest chance of having the same mutation(s) as this patient. Other more distant relatives such as cousins, aunts, uncles, and grandparents also have a chance of carrying the same mutation(s). Testing of at-risk relatives can identify those family members with the same mutation(s) who may benefit from surveillance and early intervention.

It is appropriate to offer genetic counseling to individuals with *RB1* mutations who are of reproductive age to discuss reproductive risks and options. Surveillance for retinoblastoma may be considered during the third trimester for pregnancies at risk for inheriting a *RB1* mutation.[9]

Since risks and screening recommendations begin in infancy, genetic testing for children at risk of inheriting an *RB1* mutation is appropriate.

References

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