

Understanding Your Positive *RB1* Genetic Test Result

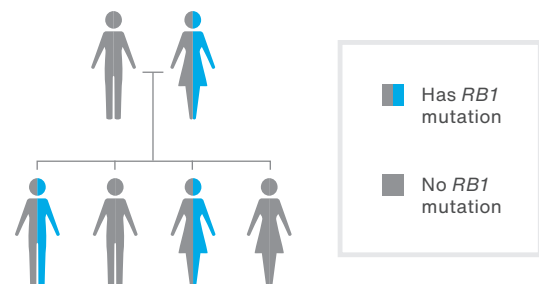
INFORMATION FOR PATIENTS WITH A PATHOGENIC MUTATION OR VARIANT, LIKELY PATHOGENIC

5 Things to know

1	<i>RB1</i> mutation	Your testing shows that you have a pathogenic mutation or a variant that is likely pathogenic in the <i>RB1</i> gene.
2	Retinoblastoma	People with <i>RB1</i> mutations have hereditary retinoblastoma.
3	Cancer risks	The risk for retinoblastoma (a form of cancer in the eye) is highest in early childhood, before 5 years. People who had retinoblastoma in childhood are at increased risk for certain soft tissue and bone cancers (sarcomas) and melanoma in adulthood. Some adults may develop sarcomas or melanoma in adulthood without a previous retinoblastoma diagnosis.
4	What you can do	Risk management decisions are very personal. There are options to detect cancer early or lower the risk to develop cancer. It is important to discuss these options with your healthcare provider and decide on a plan that works for you.
5	Family	Family members may also be at risk – they can be tested for the <i>RB1</i> mutation that was found in you. It is recommended that you share this information with your family members so they can learn more and discuss with their healthcare providers.

RB1 Mutations in the Family

There is a 50/50 random chance to pass on a *RB1* mutation to each of your children. The image to the right shows that everyone can carry and pass on these mutations, regardless of their sex at birth.



RESOURCES

- American Cancer Society [cancer.org](https://www.cancer.org)
- National Society of Genetic Counselors [nsgc.org](https://www.nsgc.org)
- Canadian Society of Genetic Counsellors [cagc-accg.ca](https://www.cagc-accg.ca)

Please discuss this information with your healthcare provider. The cancer genetics field is continuously evolving, so updates related to your *RB1* result, medical recommendations, and/or potential treatments may be available over time. This information is not meant to replace a discussion with a healthcare provider, and should not be considered or interpreted as medical advice.