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My46 Trait Profile

Retinoblastoma

Retinoblastoma is a childhood eye cancer that occurs in the retina, the light-detecting layer at the back of the eye, but can grow to fill much of the eye. It is caused by mutations in the RB1 gene which makes the retinoblastoma-associated protein.

Characteristics of Retinoblastoma

Retinoblastoma is a rare malignant (cancerous) tumor of the developing retina. It most often occurs before 5 years of age. This tumor can occur in one eye (unilateral) or both eyes (bilateral). The most common first sign of retinoblastoma is a visible whiteness in the pupil. Many parents notice this in photographs taken of their children; instead of red eye reflection they see a white eye (i.e., leukocoria). Other symptoms parents may notice are crossed eyes or eyes that do not point in the same direction, eye pain, redness, or irritation, and blindness or poor vision in the eye with a tumor.

Unilateral retinoblastoma is more common than bilateral retinoblastoma; about 60% of affected individuals have unilateral retinoblastoma. Children with retinoblastoma may or may not have a hereditary (passed through families) form of retinoblastoma. If a child has hereditary retinoblastoma it means that the gene mutation causing the retinoblastoma can be found in all of their cells; not only in their eye cells. Children with retinoblastoma in both eyes (bilateral) always have a hereditary form. Children with unilateral retinoblastoma may or may not have a hereditary form of retinoblastoma. Children with hereditary retinoblastoma are at increased risk of developing tumors outside the eye (e.g., bone cancers and melanomas).

Diagnosis/Testing

Retinoblastoma is usually diagnosed with an eye exam. Other studies such as CT, MRI or ultrasound may also be done to help with diagnosis. These studies also help the healthcare provider stage the tumor (i.e., determine how far advanced is the tumor).

Genetic testing for retinoblastoma is very important. Retinoblastoma is caused by changes or mutations to the RB1 gene. The RB1 gene is a tumor suppressor gene which means that it keeps cells from growing too fast. Mutations in the RB1 gene do not allow the retinoblastoma-associated protein to work normally and as a result, cells may grow uncontrollably. This uncontrolled growth is what can cause the tumors in affected individuals.

It is important to understand whether a child has hereditary retinoblastoma (a gene mutation of RB1 in all cells) or non-hereditary (the gene mutation only occurred in the eye). This is important for future management as well as for understanding the risk to a childs siblings and future children.

Management/Surveillance

Treatment of retinoblastoma depends on the stage of the tumor and whether there is a tumor in one or both eyes. The primary goal of treatment is preservation of life and the secondary goal is preservation of sight. Treatment can become complex and is prescribed on a case-by-case basis. Early treatment can save the life and often the vision of the patient. Treatment may include removal of the eye (enucleation), laser therapy (i.e., a procedure that destroys the tumor cells with heat) or cryotherapy (i.e. a procedure that destroys the tumor cells by freezing them), and possibly chemotherapy. A team of healthcare providers including ophthalmologists and oncologists is often required to provide the best

treatment.

If vision is impacted after treatment, it is important for patients to work with the local support of either their school or a social worker to seek services for the visually impaired.

Surveillance after initial treatment will vary based on results from genetic testing. A combination of exams under anesthesia (EUAs) and exams in the clinic are utilized to monitor for further tumor growth and new tumor growth. Exams are usually performed until 5 or 6 years of age. Individuals with hereditary RB1 mutations are at increased risk for cancers outside the eye including, sarcomas (soft tissue tumors), osteosacromas (bone tumors) and melanomas (skin tumors). A child or adult with hereditary retinoblastoma should be assessed if they complain of bone pain or lumps.

Mode of inheritance

Hereditary retinoblastoma is inherited in an autosomal dominant pattern. This means inheriting one RB1 mutation is enough for an individual to be affected. The mutation can be inherited from a parent or it can occur brand new (de novo) in a child. Most individuals with hereditary retinoblastoma have a de novo mutation.

Risk to family members

The risk to family members depends on whether the individual has hereditary or non-hereditary retinoblastoma.

If the retinoblastoma is hereditary, then the affected individuals siblings could be at risk to develop retinoblastoma. This risk depends on whether or not the affected individuals parents have a RB1 mutation. If a parent has RB1 mutation, the risk of having a child with retinoblastoma is 50% with each pregnancy.

If an individual has non-hereditary retinoblastoma, his/her siblings and future children are not at increased risk to develop retinoblastoma.

Special considerations

None

Resources

Kellogg Eye Institute: Retinoblastoma
http://www.kellogg.umich.edu/patientcare/conditions/retinoblastoma.html
Genetics Home Reference: Retinoblastoma
http://ghr.nlm.nih.gov/condition/retinoblastoma

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