What you should know about Hereditary Retinoblastoma

Retinoblastoma (RB) is a tumor of the developing retina that occurs in children. Most retinoblastomas are present before the age of 5, and some are present at birth. RBs can be present in one eye (unilateral) or in both eyes (bilateral). Approximately 60% of RBs are unilateral and 40% are bilateral. Retinoblastoma can occur randomly or be inherited. Hereditary RB is caused by mutations in the RB1 gene. Approximately 14% of children with one unilateral retinoblastoma and no family history of retinoblastoma will have a RB1 mutation. Children with multiple tumors or with one tumor and a family history of retinoblastoma have a much higher chance of having a RB1 mutation.

The risk for cancer associated with Hereditary Retinoblastoma

In addition to eye tumors, children with hereditary RB are at increased risk for other types of tumors. These include rare tumors of the pineal gland (a gland found in the brain), and cancers of bone, soft tissue, and skin. Exposure to radiation greatly increases the risk of developing secondary tumors. For this reason, it is important to identify children with hereditary RB as early as possible so that damage can be prevented.

The risks to family members

Hereditary RB is inherited in an autosomal dominant fashion. This means that the children, brothers, sisters, and parents of a person with a RB1 mutation have a 50% (1 in 2) chance of having the mutation.

Managing the risk

Surveillance in children at risk:
- Eye examination every two weeks until age three months, and every two months until age one year, and then less frequently until age four or five years.
- Brain MRI is recommended every six months until age 5 years.

Agents/circumstances to avoid:
- Avoid radiation therapy.