Parents, The First to Notice Retinoblastoma in Their Children

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1. Introduction

Retinoblastoma (RB) is a rare disease, but is the most common eye tumor in children, and the third most common tumor affecting children in the first 5 years of life. Tumor develops because of a mutation in the RB1 gene, on the 13 chromosome. This gene controls the cell growth and development. The protein produced by the RB1 gene acts as “a tumor suppressor”.

There are two forms of retinoblastoma: hereditary and non-hereditary. Patients with a hereditary retinoblastoma generally carry an alteration in one copy of the RB1 gene in all the cells of their body. If the second copy of the gene changes, retinoblastoma develops. Therefore, about 85-90% of children with hereditary retinoblastoma develop multiple tumors that affect both eyes (bilateral retinoblastoma). The remaining 10-15% of these patients develops unilateral retinoblastoma. Children with hereditary form of retinoblastoma are at an increased risk to develop tumor in the pineal gland (trilateral retinoblastoma) and possibly another cancer in the bone or muscle, later in life.

Retinoblastoma can be detected on a routine baby ophthalmologic examination, on indirect fundus exam with the pupil well dilated. In most of the cases parents notice symptoms such as: white pupil (leukocoria) instead of a normal (black) one, strabismus, red and painful eye, enlarged pupil, decreased vision.

2. Diagnosis

B-scan ultrasound –is the most common imaging test for diagnosing retinoblastoma, it is painless and doesn’t expose the child to radiation.

MRI scan provides details of the eye and the extraocular extension of retinoblastoma in the surrounding tissues, the brain and the spinal cord. It is a preferred diagnostic test as there is less radiation exposure.

CT scan can help determine the size of the tumor, the spread in the eye, and to nearby areas. CT scan shows the calcium deposits of the tumor and can be very helpful when the diagnosis is not certain.

Scintigraphy is done to show the spread of retinoblastoma to the skull or the other bones. Areas of active bones appear as “hot spots” on the scan.

3. Method

This is a two year study in the ophthalmologic and onco-pediatric department. There were 11 children recovered in both departments. In all cases children were brought to visit by the parents, because they had noticed a white reflex in the eye of their child. They were between the age of 3 to 8 years old. Children were otherwise healthy, born from a normal pregnancy. 2 of the children presented with strabismus. These symptoms had lasted from three weeks to three months before visited to the ophthalmologist.

4. Results

All patients were examined under general anesthesia with indirect ophthalmoscope, with full mydriasis; retinal calcified mass with retinal detachment was detected. On B-scan there was evidence of retinal detachments and calcifications.

Figure 1: B-scan showing retinoblastoma with calcification

Figure 2: CT–scan of the head showing no extraocular spread of retinoblastoma
Bone scintigraphy showed no spread of the tumor in the bones. Only one child had bilateral retinoblastoma, first eye enucleated three years back, then treated with chemotherapy, second eye enucleated when retinoblastoma was detected. All enucleated eyes were taken for morpho-pathologic examination, and in all cases diagnosis was retinoblastoma.

5. Conclusion

There are different treatments for retinoblastoma:

- Enucleation
- External beam radiation therapy
- Localized plaque radiation therapy
- Photocoagulation
- Cryotherapy
- Chemotherapy.

Every case of retinoblastoma is unique, and the treatment, or combination of these treatments, varies according to size, shape, and location of the tumor, whether either eyes are affected or not, and whether or not the tumor has spread (metastasis). Failure to recognize retinoblastoma at an early stage can lead to blindness and in advanced cases, death.1–3.

There is an increased risk of delayed diagnosis in younger children and in children who present with strabismus rather than leukocoria. This delay in diagnosis appears to increase the risk of local tumor invasion. Problems associated with enucleation and radiation were subsequently alleviated by use of radioactive plaque brachytherapy, 1.2.4, laser photocoagulation, 1.2.5 and cryotherapy 1.2.6. These treatments are mainly appropriate for small or recurrent tumors, often in eyes that had already undergone external beam irradiation. Therefore, the next best chance of alleviating the problem of late diagnosis of retinoblastoma, would be through education of the healthcare personnel and the parents who would possibly be first to see a child with retinoblastoma. Education of the parents is of crucial importance, because they may be the first to detect an abnormality in their children. Such education can be done by lectures or distribution of literature exhibited in paediatricians’ waiting rooms for parents to read. This can lead to an early detection of retinoblastoma, resulting in implementation of more conservative therapeutic modalities. As a result these patients can preserve some of their visual function and prevent invalidity later in life.

References