A Sporadic case of Trilateral Retinoblastoma

Dr. Aruna Fernando, Dr. Champa Banagala
Eye Hospital, Colombo

Case Presentation

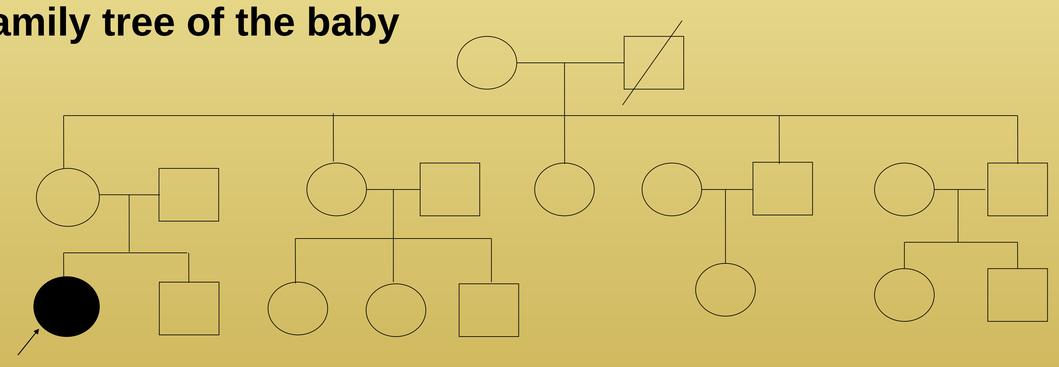
A 4 month old baby girl presented with right sided white reflex. The prenatal, natal & post-natal history was uneventful. The couple had a another healthy 10yr old boy. There was no significant family history of ocular disease.

The child had no dysmorphic features. The eyes were orthophoric. The right eye had a hazy cornea with a shallow anterior chamber. Vitreous cavity was filled with a large endophytic retinoblastoma. The left eye was normal. Right eye was enucleated and left eye was observed for development of primary tumours .

At 8 months a lesion was detected in the left eye with vitreous seedlings. Radio-imaging confirmed no cerebral metastasis or mass lesions. Local ablation as well as external beam radiotherapy & chemotherapy were administered. At 1½years the tumour showed further progression and the parents were advised on enucleation of the left eye to prevent metastasis.

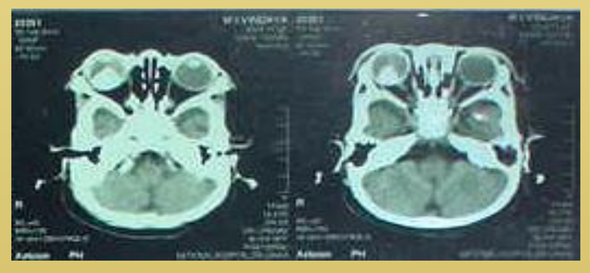
Treatment was defaulted and she presented with fits at 3½years. Neuro-imaging confirmed extensive involvement of the left orbit & a mass lesion in the pineal gland. The child passed away two months later. An autopsy was refused by the parents.

Family tree of the baby

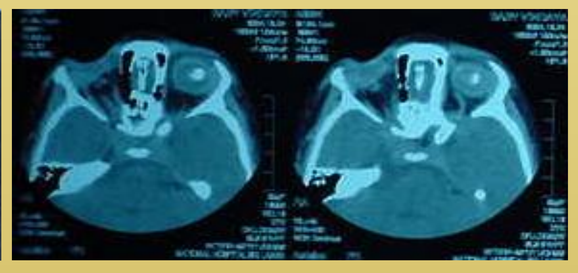


Baby at the age of 3½years

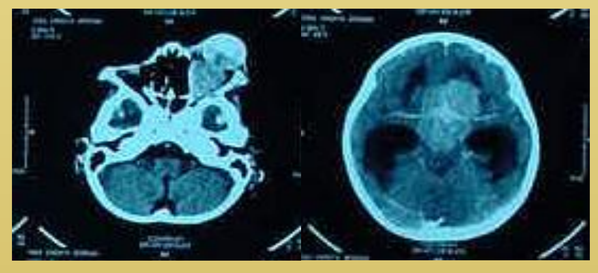
CT Scans of orbit & brain



Age-4 months
Large calcified lesion in the R/E
L/E normal



Age-8 months
Calcified lesion in L/E
No lesions in the brain



Age-3½year
Extensive involvement of the left orbit with a mass lesion in the region of the pineal gland

Histology of Right Eye

Well differentiated tumour occupying 2/3 of the vireous cavity. No involvement of the optic nerve or the choroidal vasculature.

Genetic Studies

RB gene mutations were negative in peripheral blood

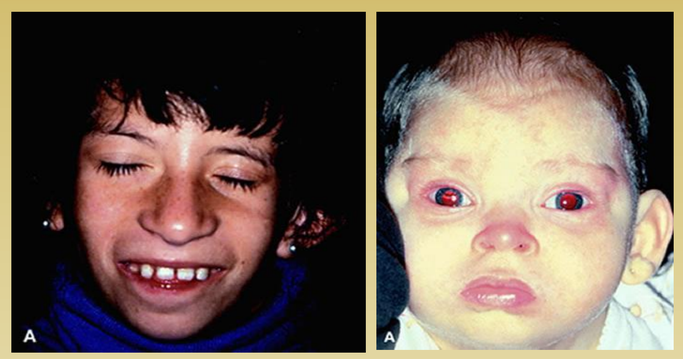
Discussion

'Trilateral retinoblastoma' designates the association of bilateral retinoblastoma with an ectopic (non-metastatic) intracranial primitive neuro-ectodermal tumor (PNET), commonly involving the pineal gland. A child may even present with an ectopic intra-cranial lesion prior to ocular involvement[1]. If treatment is not advocated once the intra-cranial tumour is diagnosed, the disease is almost always fatal within a couple of months[2].

The disease manifests due to a mutation in the tumour suppressor gene at the 13q14 locus. A majority of the mutations are gene inactivating. In 2007 alone, ten new mutations were described[4]. Mutation detection rates are around 74% for sporadic bilateral cases and 11% for sporadic unilateral cases[4]. In this patient no gene mutations were found in peripheral blood. This baby's mutation might not have been available in the currently tested markers.

Large deletions in the 13q14 locus gives rise to *trilateral retinoblastoma* with *dysmorphic features* such as microcephaly, hypertelorism, microphthalmos, epicanthus, ptosis, facial asymmetry, genital malformations, hypoplastic thumbs, toe abnormalities and mental retardation[5].

Children having 13q14 syndrome



Reference

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2. **Trilateral retinoblastoma**; Amoaku WM, Willshaw HE; *Cancer*. 1996 Aug 15;78(4):858-63
3. **Trilateral Retinoblastoma: A Meta-Analysis of Hereditary Retinoblastoma Associated With Primary Ectopic Intracranial Retinoblastoma;** Tero Kivelä; *Journal of Clinical Oncology*, Vol 17, Issue 6 (June), 1999: 1829
4. **Ten novel RB1 gene mutations in patients with retinoblastoma**; Abouzeid H, Munier FL, Thonney F, Schorderet DF. *Mol Vis* 2007 Sept 19;13:1740-5
5. **Duan's Ophthalmology 2006**, Vol-3, Chap-21,35,55