Retinoblastoma: unusual imaging findings

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Clinical History:

We present the case of a 5 year old child with proptosis on the left eye since 3 months.

Imaging Findings:

A 5 year old child with the clinical sign of strabismus on the left eye for 1 year was admitted to our hospital. The eyeball had been enlarged since 3 months. The presence of leukocoria was denied. There was no family history of the disease. Vital signs were unremarkable. Lumbar puncture and bone marrow puncture showed no tumour cell metastases. The patient underwent orbital and brain CT scan examination with contrast agent administration for determining the tumour's extension. Slice thickness 1.5 mm, interslice gap 0.6 mm. CT revealed a huge mass at the left orbital posterior eyeball and it invaded the posterior eyeball with minimal calcification component. Left optic nerves showed fusiform enlargement with smooth border (Fig 1). The mass spread into parasellar and chiasma opticum (Fig 2). Right orbita was within the normal limit. Based on the CT scan findings, the patient was diagnosed with optic nerve glioma with differential diagnoses include retinoblastoma and teratoid medulloepithelioma. The patient underwent exenteration to establish the definite diagnosis even though the tumour already spread into the cranium. Histopathology findings revealed retinoblastoma with poor differentiation (Fig 3). Post-operatively, the patient underwent combined chemotherapy and external beam radiation therapy. In the 4th cycle chemotherapy, the condition deteriorated and he became unconscious. A serial brain CT scan was then performed to evaluate the intracranial mass progression (Fig 4). Despite of aggressive chemotherapy the patient died 6 months after surgery.

Discussion:

Retinoblastoma is the most common intraocular malignancy in children. The peak incidence of retinoblastoma is 18 months old. The symptoms and signs of retinoblastoma are proptosis, leukocoria, hyperemia, buphthalmos and strabismus [1-4]. According to data from the hematolocgy department at Cipto Mangunkusumo Hospital (University of Indonesia), retinoblastoma in Indonesia is the second most common tumour after acute lymphosistic leukemia. More than 60% retinoblastoma cases in Indonesia are extraocular tumour so the most common clinical manifestation is proptotic. In this case the symptoms were strabismus and proptotic. The parents denied for leukocoria symptom. Imaging revealed a massive fusiform enlargement of the optic nerves and supra and parasellar mass. CT showed enlargement of superior orbital fissure and optic nerve. The age of the patient was older than the common age for retinoblastoma. Minimal ocular involvement with massive optic nerve and chiasmal invasion is quite exceptional combination in retinoblastoma case. Consequently, the diagnosis was glioma, but it is also uncommon that glioma optic nerves develop calcification in the eye ball. In paediatric population, the median patient age of optic
nerve glioma is 5 year old and 35% of the patients have neurofibromatosis [4]. Teratoid medulloepithelioma is
uncommon tumour of the central nervous system, typically affecting infants and young children. The tumour
develops from the non-pigmented or pigmented epithelium of ciliary body, retina and optic nerve [5]. Most
medulloepithelioma arise in childhood at a median patient age of 5 years. The optic nerve is a very unusual site of
medulloepithelioma. Teratoid medulloepithelioma can also show calcifications. The tumor most commonly develops
in the periventricular region of the cerebral hemisphere, suprasellar region, cerebellum and brainstem. On CT,
medulloepitheliomas are typically well circumscribed, iso- to slightly hypodense homogeneous mass [3]. The
enhancement is uncommon post contrast injection. On the other hand, retinoblastomas have typically hyperdense
mass on non-contrast CT and mild to moderate enhancement post contrast injection.
Since uncertainty of the diagnosis the patient underwent exentration even though the mass had already invaded the
intracranial space. To make definitive diagnosis histological examination is crucial. It is important to prove this case
because chemotherapy regimen options for retinoblastoma and other tumour are different. The histopathology
findings revealed small round cell uniform, little cytoplasmic hypercromatic with mitosis, necrotic component and
calcification without rosettes formation. These histological features were consistent with highly undifferentiated
retinoblastoma. The presence of calcification in this case is the most important clue for diagnosing retinoblastoma.
Calcification can be seen in 95% of retinoblastoma patient [3]. Even though teratoid medulloepithelioma has
calcifications the incidence of the tumour is very rare and typically lack of enhancement. Histological examination is
important to exclude the possibility of glial tumour and medulloepithelioma. Since huge mass intracranial the
prognosis of this patient was poor. He died 6 months after surgery.

**Differential Diagnosis List:** Highly undifferentiated retinoblastoma.

**Final Diagnosis:** Highly undifferentiated retinoblastoma.

**References:**

Figure 1

Description: Fig 2. The mass spread into parasellar and chiasma opticum. Origin:
Description: Fig 1a. CT: the mass was predominantly located in the posterior eye ball with fusiform and thickening of the optic nerve. Origin:
Description: Fig 1b. Post contrast CT: the mass showed enhancement.
Description: Fig 1c. Minimal intraocular calcification Origin:
Figure 3

Description: Fig.3. Histopathology revealed highly undifferentiated tumor cells and calcification within the area of necrosis. Origin:
Figure 4

Description: Fig 4. (axial) There is no improvement of the mass (post operation, chemo and EBR)

Origin:
Description: Fig 4 (sagital) Origin: