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Trilateral Retinoblastoma with Spinal Metastases

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Introduction

Trilateral retinoblastoma is a syndrome consisting of bilateral (rarely unilateral) hereditary retinoblastoma in association with an intracranial neuroblastic tumour arising usually in the pineal region, infrequently at the suprasellar or parasellar region. It can arise from either hereditary or sporadic forms of retinoblastoma. This article reports a child with uniocular retinoblastoma in association with suprasellar tumour and spinal metastases.

Retinoblastoma is the commonest primary ocular malignancy in children. Worldwide, the incidence is 1 case of retinoblastoma per 14,000 to 20,000 live births with some variation between countries.On the other hand, trilateral retinoblastoma is a rare condition. Among 141 cases of retinoblastoma seen in Hospital Kuala Lumpur (the main referral centre for retinoblastoma in Malaysia) between August 2001 and December 2009, there were only two children with trilateral disease (1.4%). It is a disease characterized by a bilateral retinoblastoma (rarely unilateral) occurred in association with an intracranial neuroblastic lesion, which usually arise in the pineal region. Less frequently, the intracranial neuroblastic lesion may be detected at the suprasellar or parasellar region as well.

Discussion

Trilateral retinoblastoma has poor prognosis for both ocular and survival outcomes. The prognosis is worse in developing countries where patients usually die of metastatic disease due to poor disease awareness and late presentation.This article reports a child with uniocular retinoblastoma in association with suprasellar tumour and spinal metastases.

Trilateral retinoblastoma is a rare condition, arising in about 3% of all the patients with retinoblastoma. It manifests either as unilateral or bilateral intraocular retinoblastoma associated with an intracranial Primitive Neuroectodermal Tumour (PNET) either in the pineal, suprasellar or parasellar region. The association of intracranial neuroblastic lesion with unilateral intraocular retinoblastoma is less common than bilateral intraocular retinoblastoma with the incidence of 0.5% and 5%-15% respectively.The intracranial lesions are considered to be independent primary retinoblastoma rather than metastatic spread as both retinal, and pineal or sellar tissue was originated from the same neural ectoderm.

Trilateral retinoblastoma can be diagnosed on the basis of eye examination, radiological imaging such as Computed Tomography (CT) scans and MRI, and then confirmed histologically. The diagnosis in our patient was challenging because he presented with concurrent retinoblastoma with intracranial and spinal lesion. Thus, the intracranial lesion needs to be differentiated between the primary tumor and metastatic lesion.

Conclusion

The diagnosis of trilateral retinoblastoma remains a challenge because he presented with concurrent retinoblastoma with intracranial and spinal tumor. Trilateral retinoblastoma was diagnosed after the HPE of ocular tumor was confirmed as retinoblastoma and the biopsy of the distal spinal cord tumor are consistent with presence of retinoblastoma cells.

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