



PRIMARY ORBITAL NEUROBLASTOMA- A RARE OCCURRENCE

Pathology

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ABSTRACT

Neuroblastoma is derived from the sympathogonia of neural crest origin. It represents as the fourth most common tumor in the first decade of life as well as second most common orbital tumor in childhood. We report a rare case of primary orbital neuroblastoma in a 6-year-old girl. She presented with painless, rapid onset proptosis of the right eye for the past four months. Computed tomography and MRI study of orbit showed enhancing soft tissue lesion measuring 26x16x15 mm into middle cranial fossa and right orbit, compressing the temporal lobe, extraocular muscles, optic nerve, causing protrusion of right optic globe. A definite diagnosis of neuroblastoma was made. Additional medical assessment comprising of chest X-ray, CECT whole abdomen showed no systemic involvement. The diagnosis of primary orbital neuroblastoma was confirmed. Primary orbital neuroblastoma even though an unusual tumor in children should be taken into account as the differential diagnosis of orbital tumors in childhood.

KEYWORDS

Primary, orbit, neuroblastoma, child

INTRODUCTION

Neuroblastoma is derived from the sympathogonia of neural crest origin. It represents as the fourth most common tumor in the first decade of life as well as second most common orbital tumor in childhood [1]. Sixty percent cases of the neuroblastoma occurs predominantly in abdomen but in only eight percent cases the tumor develops in the orbit from ciliary ganglion. Forty percent of the cases in children are older than two years of age [2].

Here, we report a case of primary orbital neuroblastoma in a 6-year-old girl.

Case report

A 6-year-old girl attended the Ophthalmic Outpatient Department at a tertiary care hospital, with painless, rapid onset proptosis of the right eye for the past four months and associated with loss of vision. On ocular examination, she had proptosis of 28 mm with lid edema and ciliary congestion with total restriction of extraocular movements. On further examination, it was fixed, not pulsating and not expandable on coughing or crying. Left eye was normal on inspection. There were no signs of congenital malformation. General examination showed no obvious abdominal mass, chest was clear and the cardiovascular system was normal.

Computed tomography of orbit showed enhancing soft tissue lesion measuring 26x16x15 mm, extending laterally to infra temporal fossa and medially into the extra conal area of orbit causing proptosis of right globe (Figure 1). MRI study of the orbit revealed intensely enhancing heterogenous lobulated altered signal intensity mass lesion extending into middle cranial fossa and right orbit, compressing the temporal lobe, extraocular muscles, optic nerve, causing protrusion of right optic globe (Figure 2).

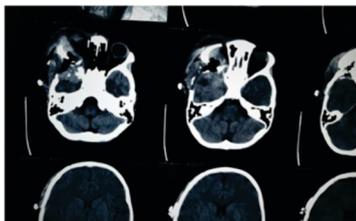


Figure 1: CT scan of Orbit shows enhancing soft tissue lesion

Fine needle aspiration cytology (FNAC) was done and the smear showed varying numbers of small blue cells with scanty cytoplasm, poorly to well-formed pseudorosettes suggestive of small blue round cell tumor (Figure 3A). Excisional biopsy (Figure 3B) showed small

round tumor cells in sheets, round to polygonal cells with high nucleocytoplasmic ratio and delicate cytoplasmic processes (Figure 3C). Vague appearance of rosettes (Homer Wright) with central fibrillary material were seen. Immunohistochemistry (IHC) staining showed positivity for synaptophysin (Figure 3D), neuron specific enolase, chromogranin. A definite diagnosis of neuroblastoma was made.

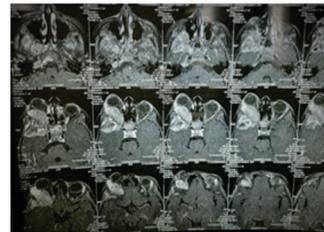


Figure 2: MRI study of Orbit shows intensely enhancing heterogenous lobulated mass lesion causing protrusion of right optic globe

Additional medical assessment comprising of chest X-ray, CECT whole abdomen showed no systemic involvement. The diagnosis of primary orbital neuroblastoma was confirmed. Complete surgical removal of the eyeball and the other contents of the eye socket was done and patient was transferred to pediatric oncology department for additional treatment and follow up.

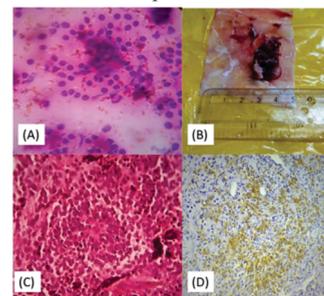


Figure 3: Orbital lesion: (A) Smear show small blue cells with scanty cytoplasm (x400, MGG) (B) Excisional biopsy (C) Section shows small round tumor cells in sheets with rosette formation (x400, H&E) (D) IHC staining for synaptophysin showing positivity (x400)

DISCUSSION

Neuroblastoma is a malignant tumour of the sympathetic nervous system developing from the primitive neuroblasts. This tumor constitutes for 8% of all childhood cancers as well as the most common

extracranial solid tumor of childhood [3]. Male to female ratio is 1.1:1 showing prevalence among males. They are predominantly metastatic to the orbit. Occasionally, they are primary lesions originating from ciliary ganglion [4].

Neuroblastoma typically disseminates through lymphatics to regional lymph nodes, mostly in the para-aortic chain in patients with abdominal tumors. Bone marrow, bone and liver are frequent sites for haematogenous spread. From three months after diagnosis, damage to the lateral orbital wall initiates periorbital ecchymosis which results in "raccoon eyes". Sixty percent of the orbital metastasis are unilateral [5]. Horner's syndrome, retinal striae, cranial nerve paralysis, anisocoria, papilledema, and nystagmus are the likely manifestations of metastatic neuroblastoma. Neuroblastoma is associated with some genetic alterations like mutations in ALK and PHOX2B as well as germline mutations in RAS pathway, TP53 and CDKN1C [6].

Microscopically, neuroblastoma is a small blue round cell tumor. The presence of neuritic processes (neuropil) which is surrounded by the neuroblasts forming Homer Wright rosettes (pseudorosette) is pathognomonic [7]. Other small blue round cell tumors like Ewing's sarcoma/primitive neuroectodermal tumor, lymphoma, rhabdomyosarcoma, retinoblastoma, desmoplastic small round cell tumor comes as differential diagnosis. Positivity for neuronal markers like synaptophysin, neuron-specific enolase, chromogranin and negative for leukocyte common antigen and desmin helps in differentiating the other small blue round cell tumors.

Cytokeratin, desmin, vimentin and neuron-specific enolase stain positive for desmoplastic small round cell tumors [8]. Rhabdomyosarcomas (RMS) are positive for desmin. Leukocyte common antigen (LCA) (CD45) express positivity for lymphoma. Ewing's sarcoma/primitive neuroectodermal tumor give diastase sensitive PAS positive reaction and express CD99.

Diagnosis of metastatic orbital neuroblastoma was excluded as there was lack of systemic symptoms and normal findings from physical, laboratory and radiologic examinations which defended our diagnosis of primary orbital neuroblastoma.

CONCLUSION

Primary orbital neuroblastoma even though an unusual tumor in children should be taken into account as the differential diagnosis of orbital tumors in childhood. The diagnosis is challenging as the histological features are shared by many small round blue cell tumors and so the immunohistochemistry can be beneficial in confining the differential diagnosis.

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