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Case Report

Radiology Section

Posterior Mediastinal Neuroblastoma with Intra-spinal Extension in an Infant: A Case Report

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ABSTRACT

Neuroblastoma is the most common extracranial solid malignancy of childhood arising from neural crest cells of the sympathetic nervous system. Although the adrenal glands are the most frequent primary site, tumours may arise anywhere along the sympathetic chain, including the posterior mediastinum. A 13-month-old female child presented with progressive upper back pain, bilateral lower limb weakness, and right-sided neck swelling for one month with a progressive nature. CT thorax revealed a heterogeneous posterior mediastinal mass extending from the superior end plate of C6 to the inferior end plate of D7 vertebral body. MRI demonstrated a 3.8×3.2×3.2 cm lesion with cranio-caudal extension from C6 to D7 (approximately 5.5 cm), resulting in severe spinal canal narrowing and cord compression. Imaging findings suggested a neurogenic tumour. Histopathological examination revealed a malignant round cell tumour - Poorly differentiated neuroblastoma. Posterior mediastinal neuroblastoma with multilevel spinal involvement is uncommon and may present with neurological symptoms such as progressive upper back pain and bilateral lower limb weakness due to spinal canal compression.

Keywords: Computed tomography, Magnetic resonance imaging, Neural foraminal extension, Paediatric thoracic tumour, Paravertebral mass, Spinal cord compression

CASE REPORT

A 13-month-old female child presented with complaints of progressively increasing upper back pain, manifested by excessive irritability, persistent crying on handling, reduced spontaneous movements, and reluctance to sit or crawl since one month.

There were progressive neurological symptoms for one month, characterised by worsening bilateral lower limb weakness and difficulty in standing/walking. Muscle power in bilateral lower limbs was reduced to Grade 2/5 proximally and 3/5 distally on the Medical Research Council (MRC) scale. Increased tone with spasticity was noted in both lower limbs. Sensory examination revealed diminished sensation below the mid-thoracic level, suggestive of a sensory level corresponding to thoracic cord compression. No bowel or bladder dysfunction was present at the time of presentation.

Normal vaginal delivery and no Neonatal Intensive Care Unit (NICU) admission were noted, and vaccinations were taken till admission date, and patient did not have any complaints of milestone regression.

The child's past medical history has no significant findings. None of the family members have similar complaints.

Complete Blood Count (CBC) revealed mild normocytic normochromic anaemia with elevated Erythrocyte Sedimentation Rate (ESR) suggestive of an inflammatory/neoplastic process [Table/Fig-1].

Parameters	Result	Reference range
Hb	9.4 g/dL	10.5-13.5 g/dL
TLC	11,800/mm^3	6,000-17,500 /mm^3
Platelet count	4.2×10^5/mm^3	1.5-4.5×10^5/mm^3
Neutrophils	62%	25-65%
Lymphocytes	32%	45-75%
ESR	42 mm/hr	<20 mm/hr

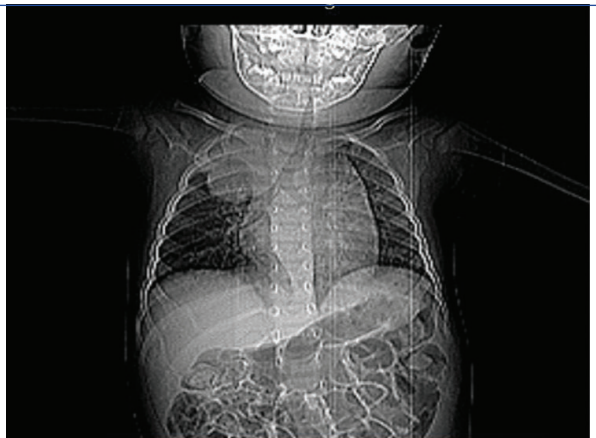
[Table/Fig-1]: Haematological parameters.
Hb: Haemoglobin; TLC: Total leucocyte count; ESR: Erythrocyte sedimentation rate

Biochemical investigations revealed markedly elevated serum Lactate Dehydrogenase (LDH) and ferritin levels along with significantly raised urinary catecholamine metabolites {Vanillylmandelic acid (VMA) and Homovanillic acid (HVA)} [Table/Fig-2].

Parameters	Result	Reference range
LDH	986 U/L	135-450 U/L
Serum Ferritin	412 ng/mL	7-140 ng/mL
Urinary VMA	34.6 mg/g creatinine	<13.6 mg/g creatinine
Urinary HVA	46.2 mg/g creatinine	<24 mg/g creatinine
NSE	25 ng/mL	<15 ng/mL

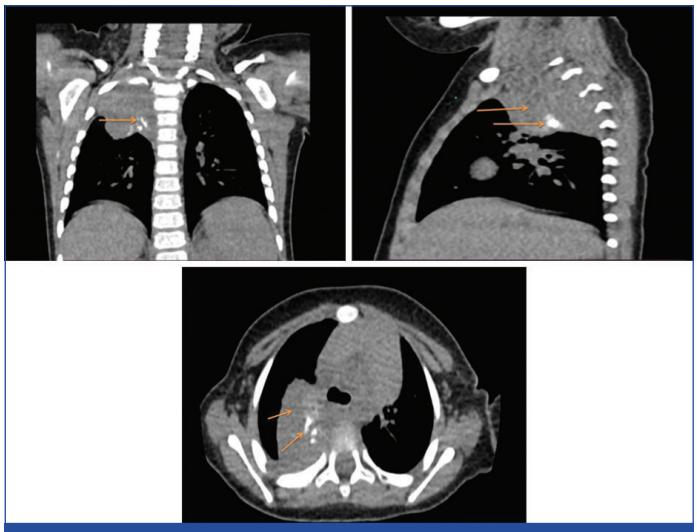
[Table/Fig-2]: Biochemical and tumour marker profile.
LDH: Lactate dehydrogenase, VMA: Vanillylmandelic Acid, HVA: Homovanillic Acid, NSE: Neuron specific enolase

Chest radiograph revealed a well-defined right paraspinal opacity in the upper lung field with mild tracheal deviation towards the contralateral side with widening of the paravertebral stripe. However, no rib or any bony involvement was noted. No evidence of pleural effusion was noted bilaterally [Table/Fig-3].



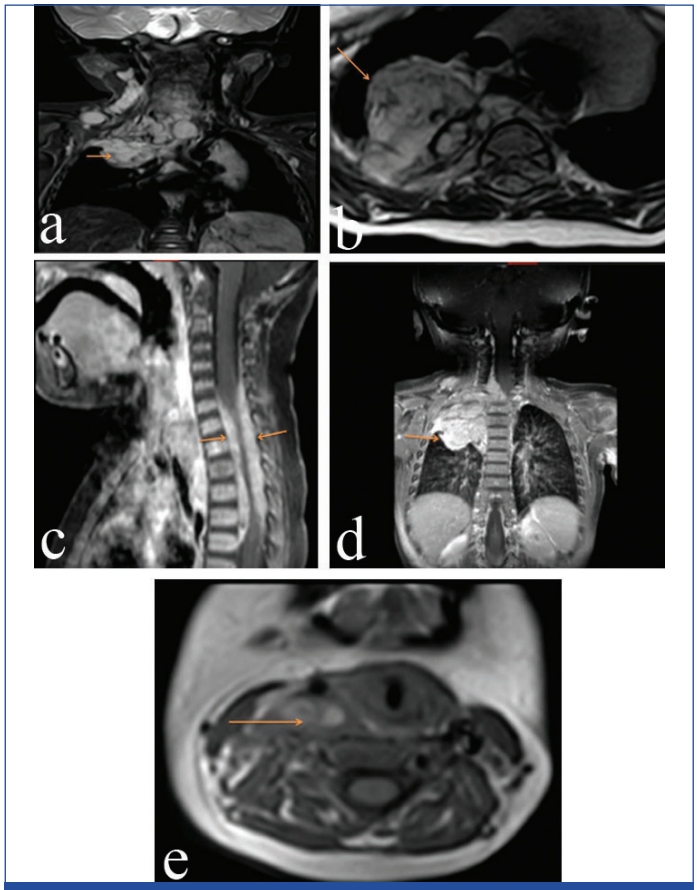
[Table/Fig-3]: Chest AP radiograph revealed a well-defined right paraspinal opacity in the upper lung field with mild tracheal deviation towards contralateral side with widening of the paravertebral stripe.

HRCT thorax demonstrated a well-defined heterogeneous soft-tissue mass in the right posterior mediastinum extending from the superior end plate of C6 to the inferior end plate of D7 vertebral levels with no foraminal widening measuring approximately 3.7×3.2×3.1 cm with internal areas of necrosis. Calcification was present. There was no evidence of any vertebral body, posterior element or rib involvement/remodeling noted [Table/Fig-4].



[Table/Fig-4]: HRCT Thorax soft-tissue window images of axial, coronal and sagittal sections demonstrate well-defined heterogeneous soft tissue mass in the right posterior mediastinum. (Below mention arrows shows calcification and mass).

MRI spine demonstrated a lobulated right posterior mediastinal paravertebral mass measuring 3.8×3.2×3.2 cm. The lesion extended cranio-caudally from the superior endplate of C6 to the inferior endplate of D7 (approximately 5.5 cm), with extension through the neural foramina into the epidural space, causing severe spinal canal narrowing and cord compression [Table/Fig-5].



[Table/Fig-5]: a) STIR sequence demonstrating hyperintense mass lesion involving posterior mediastinum; b) T2 axial sequence demonstrating hyperintense mass; c) T1 weighted sagittal contrast images demonstrating fusiform thickening with spinal canal narrowing; d) T1 weighted contrast coronal images showing post-contrast enhancement of the lesion. e) T1 axial images demonstrating extension of mass.

CT-guided core needle biopsy of the paravertebral mass was subsequently performed. The histopathology analysis with immunohistochemistry was consistent with poorly differentiated neuroblastoma with positive expression of synaptophysin and a high proliferation index (>80%) of tumour cells. Microscopically, the malignancy consisted of small round cells with a high nuclear-to-cytoplasm ratio, scant cytoplasm, and a prominent nucleolus in fibrovascular stroma surrounding blood vessels. The patient received induction chemotherapy consisting of carboplatin, etoposide, cyclophosphamide, and doxorubicin according to the institutional paediatric neuroblastoma protocol. Dexamethasone (0.5 mg/kg IV every 6 h) was administered for five days, then tapered. Dysphagia resolved on day 3, and motor strength recovered to 4+/5 on day 6. Full neurological recovery (motor strength 5/5 with normal gait) was reached by week 4, allowing discharge from hospital. Chemotherapy was well-tolerated and was associated with no more than grade 1-2 haematological side-effects, stable renal and liver functions, as well as no neuropathy and ototoxicity. MRI scan after chemotherapy was not performed during the treatment course. At 18 months follow-up, the patient was in complete clinical remission.

DISCUSSION

Neuroblastomas are the most frequent cancers during infancy. The vast majority are diagnosed in children <5 years of age, with the disease most commonly appearing between 2-3 years. In terms of location, the adrenal glands are the primary site in 35% of cases, followed by the paraspinal ganglia at about 30%, and the posterior mediastinum at approximately 20%. The less common sites include the pelvis (2-3%) and neck (1-5%), while the thymus, lungs, kidneys, stomach, and cauda equina represent extremely rare tumour locations [1].

Serum ferritin, Neuron-Specific Enolase (NSE), LDH, and urinary catecholamine metabolites- VMA and HVA- are routinely evaluated in children with suspected neuroblastoma and aid in diagnosis [2].

For neuroblastomas, multimodality imaging technique approach has to be used, with imaging modalities being divided into two: anatomical imaging (USG- usually the first screening imaging modality used in diagnosing masses; CT- after the USG flags down the primary mass, CT scans are used to visualise the fine calcifications within the mass and to get an idea about the extent of the tumour. MRI- a better imaging modality for pelvic, brain, and spinal cord imaging; functional imaging (123I – MIBG scintigraphy with SPECT and 18F-FDG PET/CT- helpful as neuroblastoma cells originate from the sympathetic nervous system) [3].

Differential diagnosis of posterior mediastinal mass includes neuroblastoma, Ganglioneuroma, schwannoma, and Neurofibroma.

Neuroblastoma predominantly occurs in children and typically appears on CT as a heterogeneous mass with areas of necrosis and calcifications, while MRI frequently shows intra-spinal extension [4,5]. Ganglioneuroma is more commonly seen in adolescents and is characterised by a well-defined, homogeneous mass on CT and a T2-weighted hyperintense lesion on MRI, with a characteristic whorled appearance due to myxoid matrix and fibrous components [6]. Schwannoma, which is usually seen in adults, appears as a well-circumscribed paravertebral mass on CT and on MRI with neural foraminal widening due to its origin from an intercostal nerve or spinal nerve root [7].

Children less than one year of age have a good prognosis, i.e., an 80-90% survival rate after five years, but those with spinal cord compression need urgent chemotherapy, surgery, and radiation to prevent neurological damage [1].

A similar case was reported by Rajparath R et al., who described an 11-month-old developmentally normal male infant presenting with irritability and progressive inability to stand or sit due to bilateral lower-limb weakness. Chest radiography demonstrated mediastinal widening, while MRI revealed a posterior mediastinal

mass with intraspinal extension. Elevated urinary VMA levels and histopathological examination confirmed poorly differentiated neuroblastoma. The child was treated with chemotherapy and remained on regular follow-up. In contrast, our patient presented with dysphagia and cervicothoracic spinal cord compression caused by a posterior mediastinal neuroblastoma extending from C6 to D7 with epidural extension [1].

Mandal KC reported two cases. One case was reported in a 10-day-old neonate presenting with paraplegia and bladder-bowel incontinence due to congenital neuroblastoma with D10-L1 intraspinal extension. Elevated urinary VMA and Fine Needle Aspiration Cytology (FNAC) confirmed the diagnosis. Although four cycles of neoadjuvant chemotherapy resulted in complete radiological resolution, the neurological deficits persisted. In contrast, our patient achieved complete neurological recovery and sustained clinical remission at 18 months following early corticosteroid therapy and multimodal chemotherapy. Case 2 was seen in a 16-month-old girl who presented with a cervical mass and paraparesis due to cervical neuroblastoma with C2-C6 intraspinal extension. Despite marked tumour regression following neoadjuvant chemotherapy, she developed severe pancytopenia, sepsis, and ultimately died of multiorgan dysfunction syndrome [8].

These cases underscore the variable clinical course of neuroblastoma with intraspinal extension and highlight the importance of early diagnosis, prompt multidisciplinary management.

CONCLUSION(S)

Posterior mediastinal neuroblastoma should be considered in the differential diagnosis of paediatric paraspinal masses. Multilevel spinal extension, as seen in this case from C6 to D7 vertebral levels, can result in significant spinal canal narrowing and neurological symptoms. CT and MRI remain indispensable imaging modalities for accurate diagnosis and assessment of intraspinal involvement.

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