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NEUROBLASTOMA IN A CHILD WITH CHRONIC CONSTIPATION – AN ATYPICAL PRESENTATION

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Abstract

Introduction: Neuroblastoma ranks as the most prevalent extracranial solid tumor in infants (1). While typically sporadic, familial occurrences have been documented (2). Originating from primitive neuroblasts of the embryonic neural crest, neuroblastoma can manifest anywhere within the sympathetic nervous system (3). The primary tumor is most frequently located in the abdomen, with approximately 65% of cases occurring there, half of which arise from the adrenal medulla. Other common sites include the neck, chest, and pelvis (4). There's no established association between prenatal or postnatal exposure to drugs, chemicals, or radiation and increased neuroblastoma incidence. Prognosis tends to worsen with advancing age. Treatment approaches encompass various modalities such as surgery, chemotherapy, radiotherapy, biotherapy, and, in select cases, careful observation alone (5). **Case report:** A 15 months female presented with complaints of abdominal distension and non-passage of stool in 3 days, with history of chronic constipation. On examination, abdomen was distended with pallor and hypertension. **Management**: Roentgenogram was suggestive of dilated fecal filled bowel loops. USG abdomen confirmed the above and revealed a retroperitoneal mass. Contrast enhanced CT abdomen was done which was suggestive of neuroblastoma. Blood investigations revealed severe anemia (Hb - 5.4 g/dl) and leukocytosis (TLC 20,450 cells/cumm - N60 L32), GBP was suggestive of microcytic hypochromic anemia with leukocytosis. Patient was managed with blood transfusion and other supportive treatment. Child was started on first cycle of chemotherapy and is doing well on follow-up. **Conclusion**: Neuroblastomas can manifest with subtle and nonspecific symptoms, potentially leading to misdiagnosis or overlooked cases. Thus, in children experiencing chronic constipation of uncertain origin, the potential for neuroblastoma should be taken into account.

Keywords: Neuroblastoma, Chronic Constipation, Extracranial Tumor.

INTRODUCTION

Neuroblastoma stands as the primary solid malignancy outside the brain during childhood, comprising 10% of all childhood tumors and contributing to 15% of pediatric cancer fatalities. (1)

The biological behavior of neuroblastoma is notably diverse; while some lesions, particularly in neonates, may regress spontaneously or evolve into less aggressive forms like ganglioneuroblastoma or ganglioma, others may progress despite intensive chemotherapy. As a tumor derived from sympathetic ganglia, neuroblastoma often produces catecholamines.

Toddlers typically present with progressive abdominal distention or discomfort, with retroperitoneal masses often encasing rather than displacing vessels and viscera. Tumor-related catecholamine production can result in symptoms like flushing, sweating, and irritability, while vasoactive intestinal polypeptide secretion may lead to secretory diarrhea. Neurologic manifestations such as opsoclonus-myoclonus, weight

loss, and anorexia may also occur. Metastases are common at diagnosis, predominantly affecting regional and distant lymph nodes, bone marrow and cortex, the orbit, liver, and occasionally the lungs, manifesting in symptoms like bone pain, proptosis, and skin lesions.

Diagnostic investigations aim to delineate tumor anatomy, size, regional invasion, metastatic spread, functional status, and histological characteristics. Elevated levels of catecholamines and their metabolites (homovanillic and vanillylmandelic acid) are typically found in spot urine samples from 90% of patients.

Serum ferritin remains a valuable marker, while neuron-specific enolase is more pertinent in tumor biopsy immunohistochemistry. Anemia may be evident in a complete blood count. Plain radiographs reveal finely stippled tumor calcifications and displacement of gas-filled bowel loops in 50% of cases. Ultrasonography confirms the tumor's solid nature and its relationship to the kidney. Prior to biopsy or resection, CT or MRI scans with arterial and venous phase contrast should be performed. Axial imaging may uncover intraspinal tumor extension or metastases. Bone marrow aspiration detects bone marrow metastases. PET scans identify recurrent or residual metastatic lesions.

Surgical resection is the primary treatment for localized neuroblastoma, with exceptions for neonates with stage MS disease and infants up to 1 year old with stage L1 small adrenal or periadrenal tumors, where careful observation may be considered. Adjuvant chemotherapy and radiotherapy are administered postoperatively based on disease stage. High-risk neuroblastoma treatment involves at least four cycles of high-dose chemotherapy followed by resection, radiation therapy, autologous stem cell rescue, and ongoing chemotherapy. The 5-year survival rate for neuroblastoma is 81%, representing a significant improvement from 30 years ago when the tumor was uniformly fatal.

CASE REPORT

A 15 months female baby presented with complaints of abdominal distension and non-passage of stool in the previous 3 days. There was on and off history of chronic constipation. On examination, patient was irritable and abdomen was distended. In addition, pallor and hypertension was present. Roentgenogram of abdomen was suggestive of dilated fecal filled bowel loops. USG abdomen confirmed the above and revealed a retroperitoneal mass.

Contrast enhanced CT abdomen was done which was suggestive of a large ill-defined heterogeneously enhancing mass lesion of size 7.0 x 6.7 x 9.6 cm with scattered areas of calcification. Superiorly the mass was extending up to diaphragm and causing cranial displacement of liver. Left lateral aspect it was causing peripheral displacement of the spleen, inferior displacement of the left kidney and lateral displacement of bowel loops. Inferiorly it was reaching up to pelvic cavity and displacing bowel loops. The abdominal aorta and proximal common iliac vessels were displaced anteriorly. The IVC, renal vein, portal vein were not visualized. Findings were suggestive of neuroblastoma (Figure 1,2,3).

Blood investigations revealed severe anemia (Hb - 5.4 g/dl) and leukocytosis (TLC 20,450 cells/cumm - N60 L32), GBP was suggestive of microcytic hypochromic anemia with leukocytosis. Liver and kidney function tests were within normal limits. Patient was managed with blood transfusion and other supportive treatment.

PET-CT was done which was suggestive of metabolically active irregular large lobulated lesion of soft tissue attenuation in retroperitoneum involving predominantly the right suprarenal region with non-separate visualization of right adrenal gland and abdominopelvic lymph nodes.

Child was started on first cycle of chemotherapy and is doing well on follow-up.

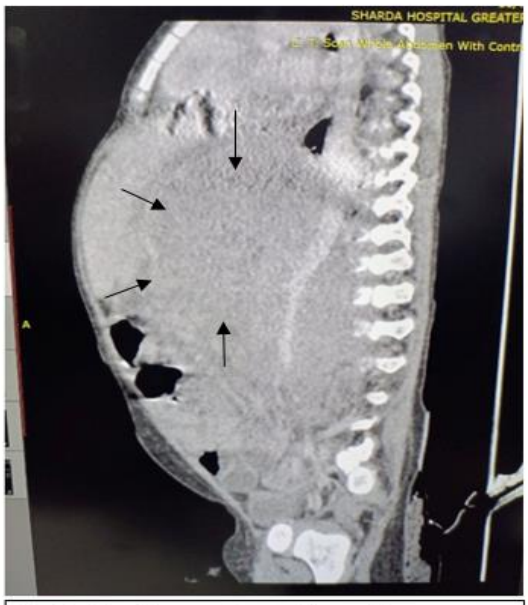

Fig 1, showing a retroperitoneal mass encasing the major vessels

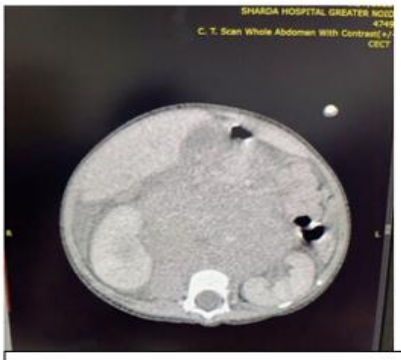

Fig 2, showing retroperitoneal mass causing a mass effect and displacing the left kidney downwards

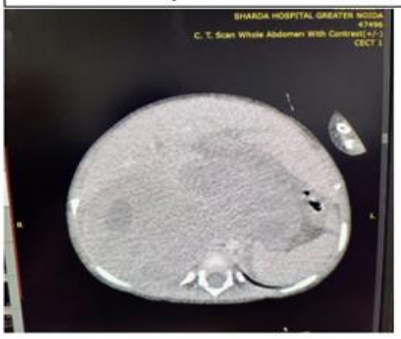

Fig 3, showing a heterogenous mass lesion with areas of scattered calcification

DISCUSSION

The signs and symptoms of neuroblastoma vary widely and encompass a diverse range, dependent on the location of the primary tumor, the presence of metastases, and any accompanying paraneoplastic syndromes. (6)

Zhang et al. reported various clinical manifestations of neuroblastomas, including neurological complications, digestive tract disorders, immune diseases and hematological disorders (7).

While numerous cases characterized by persistent diarrhea have been documented (8,9), instances of constipation presentation have been limited.

Wildhaber et al. documented two cases involving children who initially exhibited paraneoplastic syndromes attributed to ganglioneuroblastomas: the first case involved severe watery diarrhea induced by a ganglioneuroma secreting vasoactive intestinal peptide, while the second case featured untreatable constipation triggered by ganglioneuroma-produced anti-neuronal nuclear antibodies (10).

In this case, the child presented with abdominal distension and constipation and was only diagnosed with neuroblastoma incidentally after USG was suggesting a retroperitoneal mass, highlighting the importance of a thorough examination and screening.

CONCLUSION

Neuroblastomas can manifest with subtle and nonspecific symptoms, potentially leading to misdiagnosis or overlooked cases. Thus, in children experiencing chronic constipation of uncertain origin, the potential for neuroblastoma should be taken into account. Should severe constipation occur, especially in children lacking a history of gastrointestinal issues, it's wise to contemplate the presence of an undetected tumor.

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