

Case Reports

Fetal rhabdomyoma in a neonate

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Rhabdomyoma is a rare benign tumour, majority arising from the cardiac muscle. Seventy to 90% of extra cardiac rhabdomyomas are found in the head and neck region, usually within the upper aero digestive tract. We report a case of rhabdomyoma of anterior neck in a neonate. Although rhabdomyomas of posterior neck have been reported, those reported in anterior triangle are infrequent. The lesion has not recurred one year after complete excision. There are no similar reports in Indian literature.

Kew words: Newborn, Rhabdomyoma.

Introduction

The term rhabdomyoma was introduced by Zenker (1864) to indicate a benign tumour showing skeletal muscle cell with varying degree of differentiation and maturity(1). It was considered as a general diagnostic term for tumor as granular cell myoblastoma,

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alveolar soft tissue sarcoma, sarcoma botryoides, embryonal rhabdomyosarcoma, teratoma and mesenchymomas(2). Rhabdomyomas are currently defined as benign neoplasm of striated muscle tissue, consisting usually of polygonal frequently vacuolated glycogen containing cells with a fine granular deeply acidophilic cytoplasm resembling myofibril in cut section(3).

Most of the extracardiac rhabdomyomas in head and neck region in children are usually in posterior auricular region and pharynx. Anterior neck is an infrequent site. We report a case of extra cardiac rhabdomyoma of anterior neck in an infant.

Case Report

One-month-old female child presented with a swelling in the left submandibular region since birth. There was no redness or discharge and no feeding or respiratory difficulty noted. The general physical examination was unremarkable. The systemic examination did not reveal any abnormality. Local examination revealed a 8 × 6 cm swelling in left submandibular region, having bosselated surface, firm consistency, non tender, bimanually palpable and extending into the floor of mouth. The overlying skin was normal. There was no redness, scar or sinus or lymphadenopathy. X-ray soft tissue neck revealed a soft tissue mass with no calcification. Ultrasonography showed a solid lesion with heterogeneous and hyperechoic areas and no calcification. Fine needle aspiration cytology suggested a spindle cell tumor. NCCT head showed a septate cystic homogenous mass with extension superiorly behind the ramus of mandible, inferiorly till

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thoracic inlet, medially to the left para pharyngeal space displacing larynx and left carotid vessels and posteriorly reaching upto the border of sternocleidomastoid muscle. Local excision of tumor was done which revealed a well-defined solid tumor with cystic areas deep to omohyoid muscle in left sub mandibular region with no local infiltration. Histopathology of the resected tumor showed features of fetal rhabdomyoma. On histopathological examination, the tumor was composed of oval to spindle cells with vasicular nuclei. In most areas there was abundant intercellular myxoid material with few inflammatory cells (*Fig. 1*). Few cellular areas with fascicular arrangement were seen. There was no pleomorphism or mitotic activity. Occasional strap cells with abundant eosinophilic cytoplasm were seen which showed cross striations by PTAH stain (*Fig. 2*), confirming skeletal muscle differentiation. The tumor cells were positive for desmin on immunohistochemistry.

Discussion

Rhabdomyomas are rare benign neoplasms of skeletal muscle cells found more frequently in the myocardium than in the

striated muscles. The rare extra cardiac rhabdomyomas are true neoplasm and 70% occur in head and neck region. They are subdivided into adult, fetal and genital histological subtypes(4,5). The adult type of rhabdomyomas occur exclusively in the head and neck region. These are soft, coarsely lobulated tan to grey and well-circumscribed tumors ranging in size from 0.5 to 6.0 cm. Age of the patients range from 16 to 82 years (mean age 52 years). There is a marked male predominance of almost 5:1. A clonal balanced translocation has been found in Chromosome 15 & 17 in the head and neck rhabdomyoma(6). Majority of them require local excision with few local recurrence. Each recurrence is usually treated by local excision.

Fetal rhabdomyomas are benign tumors with skeletal muscle differentiation that have a propensity to occur in head and neck region. They are composed of elongated spindle shaped skeletal muscle elements in varying stages of differentiation. They are subdivided into two histological sub types "classic" immature histology and others with more prominent rhabdomyoblastic maturation "intermediate"

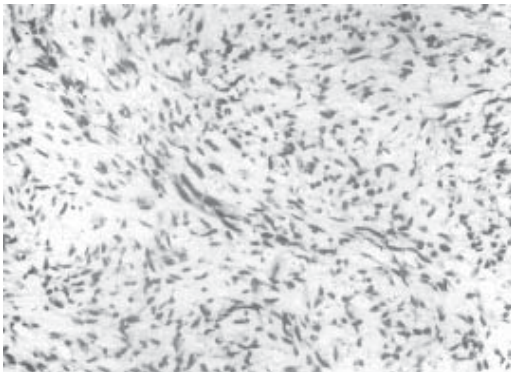


Fig. 1. Photomicrograph of the tumor to show spindle cells in a loose Myxoid background (H. & E. × 140).

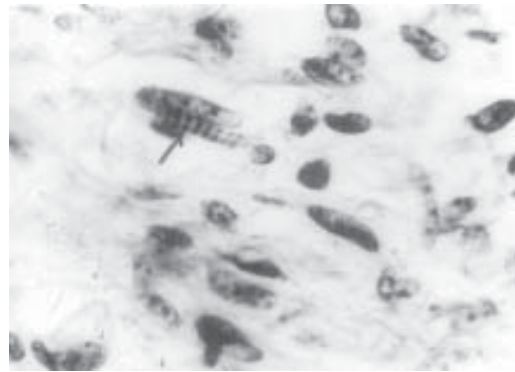


Fig. 2. Photomicrograph to show cross striations (arrow) in the tumor cell (PTAH × 1360).

histology(7). However, several tumors show overlapping features between classic and intermediate groups suggesting that these are more likely a single group with a spectrum of rhabdomyoblastic differentiation(7).

Fetal rhabdomyomas usually present as solitary mass within the soft tissue or the mucosal areas of head and neck. They may also present with hoarseness, dysphasia or respiratory distress. The most common sites are post auricular region, pre auricular region, or face, followed by nasopharynx and oral cavity. The differential diagnosis includes rhabdomyosarcomas (spindle cell variant) and benign hamartomatous lesions, such as neuro-muscular hamartomas and rhabdomyomatous mesenchymal hamartomas of the skin(8,9). Distinction from spindle cell variant of rhabdomyosarcoma and other embryonal rhabdomyosarcomas may be difficult. Unlike rhabdomyosarcomas, which has infiltrative margins and invades normal tissues, fetal rhabdomyomas are well circumscribed and do not invade and destroy adjacent soft tissue. Histologically foetal rhabdomyomas rarely show areas of necrosis, and unlike rhabdomyosarcomas they lack hypercellularity, a "cambium layer" typical of botryoid rhabdomyosarcomas, nuclear atypia, abnormally distributed chromatin and absent or low mitotic activity(10).

The subgroups of extra cardiac rhabdomyomas can further be characterized by immunocyto-chemical studies staining the myoglobin desmin vimentin muscle specific actin. Desmin is the most reliable marker for cells with skeletal or smooth muscle differentiation as it is present in both primitive and mature cells. Vimentin is present in primitive cells and myoglobin is present in mature cells(11).

The possibility of fetal rhabdomyoma

should always be considered in differential diagnosis of a cervical swelling especially in a neonate.

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Hyperammonemia with Citrullinemia

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Two cases of hyperammonemia with elevated citrulline are reported, one resulting from a deficiency of pyruvate carboxylase and the other from a partial deficiency of argininosuccinate synthetase. Diagnosis was based on clinical, biochemical and amino acid profiles. The utility of amino acid determinations in hyperammonemia suspected to underlie an inborn error of metabolism is emphasized.

Key words: Citrulline, Hyperammonemia.

Inborn errors of metabolism (IEM) are of concern in India, the spectrum being wide, varied and poorly diagnosed. Population

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based studies indicate tyrosinemia, maple syrup urine disease and phenylketonuria to be the commonest inborn errors of amino acid metabolism among newborns in India(1). Amino acid disorders seen in a tertiary care hospital would be more varied, as they would include sick children. We report 2 cases of citrullinemia with hyperammonemia which represent the disruption of different metabolic pathways.

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Patient 1. A neonate presented at 20 hours of age with poor feeding and respiratory distress. He was born to consanguineous parents and two previous siblings had died in the neonatal period with similar complaints. There was no history of birth asphyxia or any risk factors for sepsis. The blood counts, blood sugar, electrolytes and chest X-ray were normal and blood culture was sterile. On examination he was in severe respiratory distress with weak peripheral pulses and a capillary refill time >4 seconds. Arterial blood gas revealed severe metabolic acidosis with an anion gap of 35. Serum urea was 37 mg%, plasma ammonia 294 µg/dL (normal 20-80 µg/dL) and serum lactate 4.5 mM (normal 0.3-1.3mM). Urine was negative for reducing substances and ketones. Amino acid profiles determined by high performance liquid chromatography, revealed raised levels of citrulline (440 moles/L), proline (410 moles/L) and lysine (680