Fetal Rhabdomyoma

A Case Report with the Diagnosis Suggested by Intraoperative Cytology

Ammar C. Al Rikabi, M.D., M.R.C.Path., F.I.A.C., Turki Al Kharfy, M.B.B.S, F.R.C.P.C., F.A.A.P., Mohamed Omar Al Sohaibani, M.D., F.R.C.A.P., and Asal Izzdien Al Samarrai, M.D., F.R.C.S.

BACKGROUND: Fetal rhabdomyoma is a relatively rare tumor that occurs mainly in the head and neck of toddlers. A similar lesion can occasionally be found in the adult female genital tract. Increased cellularity may be seen in the fetal type and can lead to confusion with the well-differentiated types of rhabdomyosarcoma.

CASE: An 8-day-old infant presented with a unilateral, right-sided, postauricular mass. Intraoperative cytology of the mass showed numerous cells with spindled nuclei in a background of myxoid material. A provisional diagnosis of fetal rhabdomyoma was made and subsequently confirmed by histopathologic examination.

CONCLUSION: In neonates presenting with cervical swelling, the possibility of fetal rhabdomyoma should always be considered and confirmed by both intraoperative cytology and histopathologic examination of the resected tumor. (Acta Cytol 1996;40:786–788)

Keywords: rhabdomyoma, fetal diseases.

In a neonate presenting with a cervical swelling in the preauricular or postauricular region, fetal rhabdomyoma should always be investigated.

Fetal rhabdomyoma is a tumor of striated muscle origin that is relatively rare and seen almost exclusively in the head and neck area (particularly the retroauricular region) in children under 3 years of age. A similar lesion can also occur in the vulvovaginal area in middle-aged women. We report a case of fetal rhabdomyoma in which the diagnosis was suggested by intraoperative cytology. The diagnosis was later confirmed by paraffin sections and special and immunohistochemical stains.

Case Report

An 8-day-old female infant was referred to the Department of Pediatrics, King Khalid University Hospital, with a history of a right-sided cervical swelling present since birth. There were no other associated symptoms, and the antenatal period was uneventful. On examination, a hard mass, approximately 7 cm in diameter, was seen in the right postauricular region. The differential diagnosis at

From the Departments of Pathology, Paediatrics and Surgery, King Khalid University Hospital, Riyadh, Saudi Arabia.

Dr. Al Rikabi is Assistant Professor, Department of Pathology.

Dr. Al Kharfy is Assistant Professor, Department of Paediatrics.

Dr. Al Sohaibani is Associate Professor, Department of Pathology.

Dr. Al Samarrai is Professor, Department of Surgery.

Address reprint requests to: Ammar C. Al Rikabi, M.D., M.R.C.Path., F.I.A.C., Department of Pathology, King Saud University, P.O. Box 7925 (32), Riyadh 11461, Saudi Arabia.

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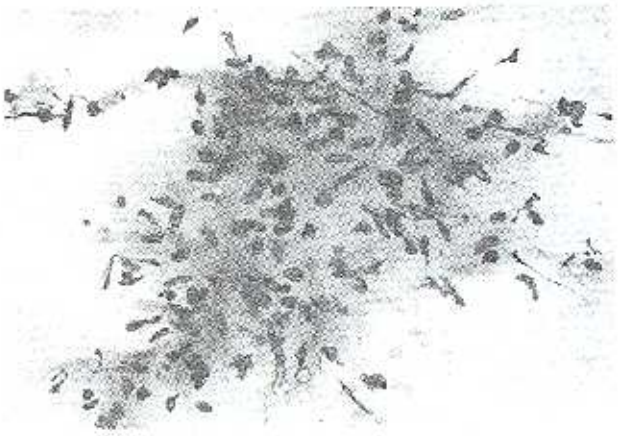

Figure 1 Intraoperative cytology of fetal rhabdomyoma. Spindle and oval nuclei in a background of myxoid stroma (Diff-Quik, × 400).

this stage included teratoma, lipoma and cystic hygroma. The mass was subsequently excised and sent unfixed for cytologic and histopathologic assessment.

The specimen consisted of a well-circumscribed, oval mass of pale, firm tissue measuring 7 × 5.5 × 3 cm. On sectioning, the cut surface was pale, homogeneous and slightly mucoid. Imprints and scrapings were made from the cut surface of the lesion, and the smears were stained using the Diff-Quik method. Cytologic examination showed a blood-stained, cellular specimen consisting of numerous cells with spindle- and oval-shaped nuclei in a background of myxoid material (Figure 1). A few scattered mononuclear inflammatory cells were also present.

There was no evidence of cellular pleomorphism, and mitoses were absent. Careful search showed absence of glial and fat cells. Based on these findings as well as the age of the patient and location of the lesion, a provisional diagnosis of benign spindle cell tumor, most probably a fetal rhabdomyoma, was made.

Histologically the tumor consisted of numerous mesenchymal cells with spindle-shaped, bland nuclei. Long cytoplasmic processes were visible in some cells (Figure 2). Cross-striations were seen in a number of cells in sections stained with phosphotungstic acid hematoxylin (Figure 3). These cells were more frequent in the large and better-differentiated fibers located in the peripheral parts of the tumor. The tumor cells showed strong positive staining with desmin and vimentin and negative staining with S-100 protein. The myxoid ground substance of the tumor consisted of Alcian blue positive acid mucopolysaccharides. Based on the above histologic features and on special and immunohistochemical staining, the cytologic diagnosis of fetal rhabdomyoma was confirmed.

Discussion

Fetal rhabdomyoma was first described in 1972 by Dehner and Enzinger,[1] who suggested that the lesion may be a hamartoma and not a true cytoplasm. The lesion is now regarded, however, by most authorities as a rare benign tumor with skeletal muscle differentiation.

Fetal rhabdomyoma occurs mainly in the head

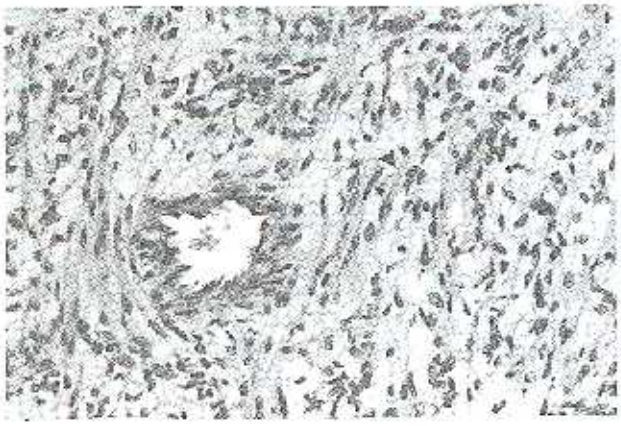

Figure 2 Spindle-shaped cells with long cytoplasmic processes (arrowhead). A blood vessel is visible on the left (hematoxylin and eosin, × 400).

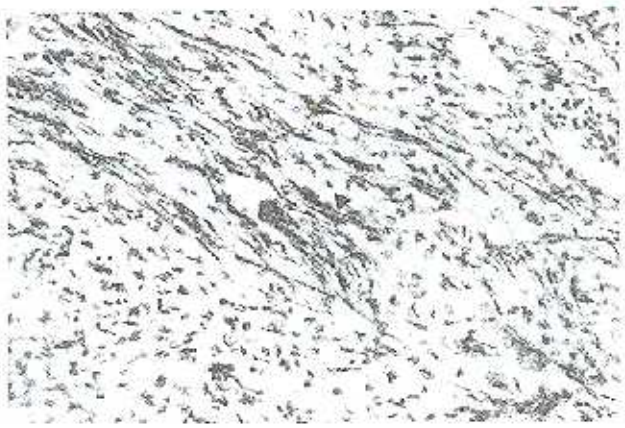

Figure 3 Cross-striations (arrowhead) are visible within the cytoplasmic processes of some cells (P.T.A.H., × 400).

and neck of toddlers, although it can be seen in older age groups.[2] A similar lesion is found rarely in the posterior urethra of children.[3] The tumor may also be associated with the nevoid basal cell carcinoma syndrome.[4]

In a study done by Kapadia et al[2] on 24 proven cases of fetal rhabdomyoma, the median patient age was 4.5 years and the male:female ratio 2.3:1. The most common soft tissue site was the face or preauricular region (7 cases), followed by the postauricular region (5 cases) and neck (4 cases). Cases of fetal rhabdomyoma have also been found in the parotid gland,[5] tongue,[6] larynx[7] and orbit.[8]

This case was an example of the classic, or immature, form of fetal rhabdomyoma, which showed, on intraoperative cytology, a mixture of undifferentiated spindle and oval cells in an abundant fibromyxoid stroma. The skeletal muscle origin of the neoplasm was confirmed by the presence of delicate cytoplasmic cross-striations and positive staining with desmin. In the intermediate form of fetal rhabdomyoma, rhabdomyoblasts showing abundant eosinophilic cytoplasm are usually present.[2] To the best of our knowledge, this is the first confirmed case report that describes the cytologic features of the immature form of fetal rhabdomyoma.

Fetal rhabdomyoma should be differentiated from the spindle cell variant of rhabdomyosarcoma. Unlike rhabdomyosarcoma, fetal rhabdomyoma usually shows well-demarcated margins with an absence of necrosis, cambium layer, nuclear pleomorphism and mitoses.

In a neonate presenting with a cervical swelling in the preauricular region or postauricular region, fetal rhabdomyoma should always be investigated. The pathologist should be familiar with the cytologic and histologic features of this rare neoplasm, which in some cases mimics the spindle cell variant of rhabdomyosarcoma.

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