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Foetal rhabdomyoma with fine-needle aspirate cytology correlation

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ABSTRACT

A case of intermediate form of foetal rhabdomyoma with cytological correlation is reported in a ten-year-old girl who presented with a lump in the right neck region. Fine-needle aspirate of the lump was performed. Cytological findings were that of spindled cells and rhabdomyoblasts with abundant eosinophilic cytoplasm. The lesion was subsequently excised. Histology showed a well-circumscribed cellular lesion composed of oval- to spindle-shaped cells. There were interspersed immature skeletal muscle cells with uniform nuclei and eosinophilic tapered cytoplasm and ganglion-like rhabdomyoblasts. No marked cellular atypia or prominent mitoses was noted. Immunohistochemically, the tumour cells showed positivity for muscle specific actin, myoglobin and myogenin. There was focal positivity for desmin. The patient showed no evidence of local recurrence or metastasis after a 32-month follow-up. This is believed to be the first case report of cytological findings in an intermediate form of foetal rhabdomyoma.

Keywords: foetal rhabdomyoma, fine-needle aspiration cytology, intermediate foetal rhabdomyoma, neck mass, rhabdomyoma

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INTRODUCTION

Foetal rhabdomyoma is a rare benign tumour showing immature skeletal muscle differentiation with a predilection for the head and neck region. Two variants, the classic and intermediate forms, have been described.[1] To the best of our knowledge, there has only been one reported case of classic foetal rhabdomyoma with cytological correlation by al Rikabi et al, which involved intraoperative cytology.[2] Cytological findings in the intermediate form of foetal rhabdomyoma have not been previously described. We report a case of intermediate foetal rhabdomyoma with fine-needle aspirate (FNA) cytological correlation.

CASE REPORT

The patient was a ten-year-old girl who presented with a lump in the right neck region. An FNA was performed on the lesion. Smears were prepared; some were fixed in ethanol for Papanicolaou (Pap) staining, and others were air-dried and stained using the Diff-Quik method. The cytological findings were that of a low-cell yield containing cells with spindle- and oval-shaped nuclei with scanty to small amount of cytoplasm (Fig. 1). There were scattered larger cells containing abundant dense eosinophilic cytoplasm and oval, peripherally-located nuclei. Some of these cells show multinucleation and nuclear palisading (Fig. 2). Few mononuclear inflammatory cells and neutrophils were present. No obvious marked cytological atypia was noted.

The lesion was later excised. The operative findings were of a 3-cm lobular lesion on the trapezius which was thought to be a lymph node. Macroscopically, the

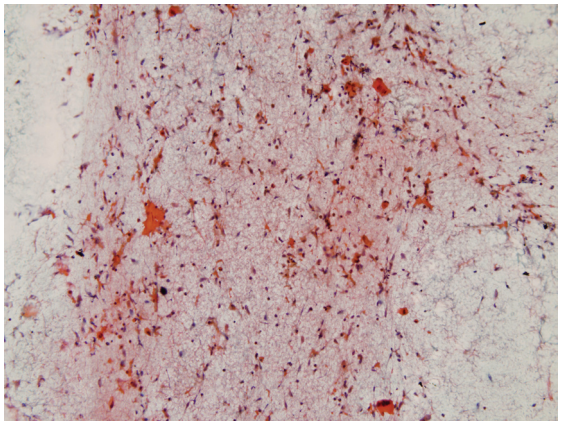

Fig. 1 Spindled cells with scanty amount of cytoplasm and larger cells containing abundant, dense eosinophilic cytoplasm and oval, peripherally-located nuclei (Pap stain, × 200).

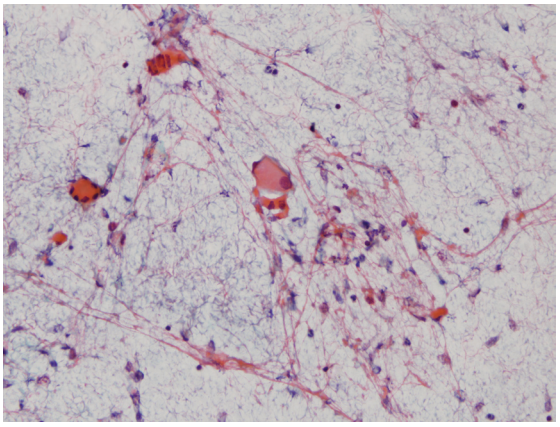

Fig. 2 Some cells show multinucleation and nuclear palisading. No obvious marked cytological atypia (Pap stain, × 400).

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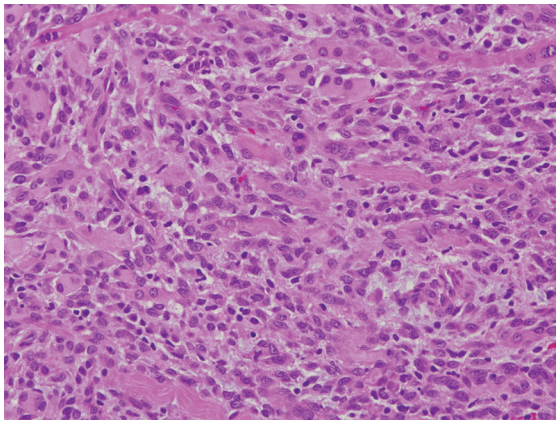

Fig. 3 Photomicrograph shows a cellular lesion composed of oval- to spindle-shaped cells with interspersed immature skeletal muscle cells showing cytoplasmic cross-striations, uniform nuclei and eosinophilic tapered cytoplasm. Some ganglion-like rhabdomyoblasts with large vesicular nuclei and prominent nucleoli are noted (Haematoxylin & eosin, × 400).

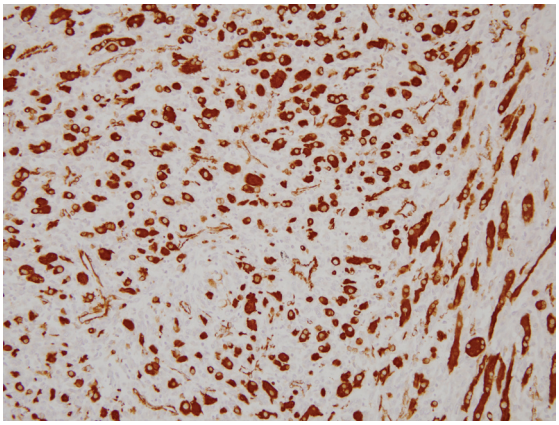

Fig. 4 Tumour cells are positive for muscle-specific actin on immunostaining (muscle-specific actin immunostain, × 100).

specimen consisted of an oval mass measuring 3 cm × 2.5 cm × 1.5 cm. Histological findings were that of a well-circumscribed cellular lesion composed of oval- to spindle-shaped cells with indistinct cytoplasm set in a variably myxoid stroma (Fig. 3). There were interspersed immature skeletal muscle cells with uniform nuclei and eosinophilic tapered cytoplasm. Some tendency to nuclear palisading was appreciated. Occasional cells showed cytoplasmic cross-striations. Some ganglion-like rhabdomyoblasts with large vesicular nuclei and prominent nucleoli were noted. No marked cellular atypia or prominent mitotic figures were present. On immunohistochemistry, the tumour cells stained positively for muscle-specific actin (Fig. 4), myoglobin and myogenin. There was focal positivity for desmin. The tumour cells showed a low proliferative index by MIB1. The appearances are consistent with an intermediate form of foetal rhabdomyoma. The tumour was present at the resection margins. However, at follow-up 32 months postoperation, the patient was alive and well, with no evidence of tumour recurrence.

DISCUSSION

Foetal rhabdomyoma was first described by Dehner et al in 1972 in a study analysing nine cases.[3] Since then, a few other series and case reports have further characterised this rare entity.[1,4,5] The majority of foetal rhabdomyomas occur in the soft tissue or mucosa of the head and neck region. Two variants of foetal rhabdomyoma have been described: the classic and intermediate forms.[1] The classic foetal rhabdomyoma has a predilection for the postauricular soft tissue, and the intermediate variant occurs more often in soft tissue or mucosal sites of the head and neck region. In one series of 24 head and neck foetal rhabdomyomas by Kapadia et al, the median age at diagnosis was 4.5 years (range 3 days to 58 years), and a male to female ratio of 2.3:1.[1] Half occurred in patients < 3 years of age. The median tumour size was 3.0 (range 1.0–12.5) cm, and presented as a well-defined, solitary mass.[1] Macroscopically, it had a soft, grey-white to tan-pink glistening, mucoid cut surface. Cases of foetal rhabdomyoma have been reported in patients with nevoid basal cell carcinoma syndrome.[6]

Histologically, the classic foetal rhabdomyoma consists predominantly of bland primitive spindled cells associated with elongated skeletal muscle cells reminiscent of foetal myotubules, displaying occasional cross striations. These are haphazardly-arranged in a fibromyxoid stroma. The intermediate forms show a greater degree and a greater number of cells with skeletal muscle differentiation as well as a variety of distinctive cytological and architectural features. These include the presence of large, ganglion cell-like rhabdomyoblasts with vesicular nuclei and prominent nucleoli, interlacing ribbon or strap-like rhabdomyoblasts, broad bundles of spindled rhabdomyoblasts simulating smooth muscle, plexiform pattern, focal intimate association with peripheral nerves, and rare areas of fibroblastic proliferation.[1] The cells show a slight tendency towards nuclear palisading.[4] Most cases were devoid of mitosis, but in a series, five out of 24 tumours had one to 14 mitoses/50 high-power field.[1] Marked nuclear atypia were uniformly absent. Immunohistochemically, foetal rhabdomyomas typically stained for myoglobin, desmin, and muscle-specific actin, with focal or rare staining for vimentin, smooth muscle actin, S-100 protein, glial fibrillary acidic proteins and Leu-7.[1] Cytokeratin, epithelial membrane antigen and CD68 antigen were negative.[1] Electron microscopy demonstrates skeletal muscle differentiation with rhabdomyoblasts containing

thick and thin myofilaments with Z-bands and glycogen within the cytoplasm.[1,3,5]

Recommended treatment is complete excision. Rare local tumour recurrence has been reported, normally attributed to incomplete removal.[4,7] No instance of aggressive local tumour growth or metastasis has been documented. Foetal rhabdomyoma can be confused with an embryonal rhabdomyosarcoma histologically. Features that favour the former are superficial location, circumscription, lack of infiltrative margin or destruction of adjacent soft tissue, evidence of cellular maturation, lack of cellular atypia and paucity of mitotic figures.[1,3,4]

Only one paper was found that described cytological findings in foetal rhabdomyoma, where it described an intraoperative cytology with imprints and scrapings made from the cut surface of the lesion. The cytological findings were that of numerous cells with spindled nuclei in a background of myxoid material. A provisional diagnosis of foetal rhabdomyoma was made, and subsequent histopathological examination confirmed a classic form of foetal rhabdomyoma.[2] The case described in this article had a diagnostic FNA done which also revealed spindled cells. Additionally, there were larger rhabdomyoblasts with abundant dense eosinophilic cytoplasm. There was some nuclear palisading. These findings were recapitulated in the histological findings of the resected specimen. The cytological findings in this case were initially interpreted as reactive or reparative in nature. A recommendation for review and excision if the lesion persists was offered. The lesion was subsequently excised and the histological findings were that of an intermediate form of foetal rhabdomyoma.

The cytological findings can mimic rhabdomyosarcoma or rhabdomyoma. Cytological findings of rhabdomyosarcoma, however, usually reveal small round cells with hyperchromatic, pleomorphic nuclei and scant cytoplasm. Rhabdomyoblasts are also identified.[8] Adult rhabdomyoma typically contains cohesive clusters of round to polygonal cells, with abundant eosinophilic granular cytoplasm and small uniform peripherally-located nuclei.[9] Cytoplasmic cross striations and crystalline inclusions may be identified. Spindled cells, however, are usually not conspicuous. In summary, one needs to be aware of foetal rhabdomyoma, a rare and benign tumour with a predilection for the head and neck region. FNA cytological material from such a tumour shows spindled cells and rhabdomyoblasts. This needs to be differentiated from rhabdomyosarcoma.

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