Foetal rhabdomyoma with fine-needle aspirate cytology correlation

Wan W K, Sng T Y, Goh H K, Hwang S G

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

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. 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. 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 lesion 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.
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. Since then, a few
other series and case reports have further characterised
this rare entity. 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. 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. 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. 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.

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. The cells show a slight tendency towards
nuclear palisading. Most cases were devoid of mitosis,
but in a series, five out of 24 tumours had one to 14
mitoses/50 high-power field. 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. Cytokeratin, epithelial
membrane antigen and CD68 antigen were negative.
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.\(^6\) 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.

**REFERENCES**