Rare disease

Congenital infantile myofibroma causing intrauterine death in a twin

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Summary

While infantile myofibromatosis is the most common mesenchymal tumour of infancy, only around 300 cases have been reported. The authors report a 33-year-old para 1 with an uncomplicated, dichorionic diamniotic twin pregnancy who was diagnosed with an intrauterine death of one twin at 36+5 weeks gestation. At caesarean section, a macerated male stillborn weighing 2.72 kg was delivered. Postmortem examination revealed a pedunculated lesion attached to the left shoulder and underlying muscle consistent with a congenital myofibroma. The cause of death was postulated to be haemorrhage from the tumour surface causing fetal anaemia.

BACKGROUND

We report only the second case of this benign neoplasm causing fetal demise and the first in a twin pregnancy. Our case is also unique because if our hypothesis is correct, it is the first case of fetal demise where visceral involvement has not been a factor.

CASE PRESENTATION

A 33-year-old para 1 with an uncomplicated DCDA twin pregnancy presented at 36+5 weeks with tightenings. An ultrasound scan performed 2 days earlier had shown normal growth, liquor volume and umbilical artery dopplers in both twins, although views had been limited due to maternal body habitus. At presentation, an intrauterine death of the presenting breech twin was diagnosed on ultrasound. At caesarean section, a macerated male stillborn weighing 2.72 kg and another male in good condition weighing 3.04 kg were delivered. The amniotic fluid around the dead twin was found to be lightly blood stained at the time of the caesarean section and a large lesion was seen on the left shoulder which had not been picked up on previous ultrasound scans.

OUTCOME AND FOLLOW-UP

Postmortem examination revealed a 8×6×6 cm firm, pedunculated lesion attached to the left shoulder and underlying muscle (figure 1). Microscopic and immunohistological examination showed a spindle-cell tumour with haemangiopericytoma-like areas with CD34 and smooth muscle actin positivity, suggesting congenital myofibroma (figures 2–4). There was surface ulceration (figure 5). Other features noted were fetal pallor, a marked increase in nucleated cells in the circulation and increased liver haemopoiesis, findings suggestive of fetal anaemia. There was no cardiomegaly present. No other cause was found so it is suggested that there was loss of blood from

the tumour surface and this may have contributed to fetal demise.

DISCUSSION

Infantile myofibromatosis (IM) is usually congenital with the median age of presentation being 3 months.¹ There are three types – solitary, multicentric or generalised. The solitary form is the most common with a male predominance. It frequently presents as a subcutaneous nodule in the skin or tissue of the head and neck but can also take the form of ulcers, pedunculated lesions or resemble haemangiomas. Lesions can also commonly appear in muscle and bone.

IM has an excellent prognosis with the majority spontaneously regressing without the need for treatment. This regression is thought to be secondary either to apoptosis or overgrowth of the vascular supply.^{2–3} Visceral involvement is rare and carries a high reported mortality rate of over 70%.⁴ Surgical excision, radiotherapy, local glucocorticoid injections and chemotherapy are reserved for those tumours that are affecting organ function.^{1–5–6}

Most cases are sporadic, but an autosomal dominant pattern with variable penetrance and an autosomal recessive inheritance have both been postulated for familial cases.⁷

Prenatal detection with ultrasound is difficult with only three cases previously reported.⁸ Disruption to the sonographic appearance of organs, presence of an abdominal mass and abnormal or limited fetal movements between 30 and 32 weeks gestation may point towards IM.⁹

Diagnosis is usually made on the histological appearance of well-circumscribed nodules showing zonal architecture, with a central area of vascular proliferation in a haemangiopericytoma-like pattern surrounded by fascicles of fibroblastic and myofibroblastic cells resembling smooth muscle.¹⁰

Figure 1 Macroscopic view of congenital myofibroma attached to the left shoulder of the foetus.

Figure 2 H&E stain of the tumour showing short fascicles of oval or slightly spindled cells with abundant eosinophilic cytoplasm.

Figure 3 Immunohistology showing the tumour cells are CD34 positive.

Figure 4 Immunohistology showing that the tumour cells are smooth muscle actin positive.

Figure 5 H&E stain showing tumour ulceration and necrosis at the surface with some areas of fibrin.

Learning points

- Infantile myofibromatosis, while rare, is the most common mesenchymal tumour of infancy.
- Prenatal diagnosis with ultrasound is difficult, but disruption to the appearance of organs, presence of abdominal mass or abnormal or limited fetal movements may point towards the diagnosis.
- Mortality is rare and visceral involvement accounts for all cases of intrauterine death up until now.
- Haemorrhage from the tumour surface may be another mechanism for in utero demise as in this case.

Competing interests None.

Patient consent Obtained.

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