A rare case of neonatal proptosis

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Abstract

Neonatal proptosis is an uncommon condition which can be a cause of major concern to family members and healthcare professionals alike. Neoplastic, cystic, vascular and miscellaneous causes occur. Early diagnosis through antenatal imaging may improve outcomes. We present a very rare case of primary congenital fibrosarcoma of the orbit presenting with severe neonatal proptosis.

Key words: orbit, proptosis, fibrosarcoma, neonatal, congenital

Funding: No funding was received for this case study.

Conflict of interest: None of the authors had any conflict of interest in respect of this case study.

Introduction

Although uncommon, proptosis at birth is a cause of immediate concern to family members as well as healthcare professionals. The condition has significant complications – such as loss of vision, deformation and potentially death. This places great importance on timeous diagnosis and management.[1] There is no recorded incidence for neonatal proptosis even though the condition is well documented. The causes of neonatal proptosis can be grouped into five broad categories: benign neoplastic, malignant neoplastic, cystic, vascular and miscellaneous. Antenatal ultrasonography can help identify these conditions early and allow for expedited management.[2,3]

Case report

A neonate presented to the Red Cross War Memorial Children's Hospital on day one of life. The child was found to have severe left proptosis on delivery by difficult elective Caesarean section. The mother had been admitted to hospital four weeks prior to delivery after suffering blunt trauma to her abdomen during an alleged assault by her partner. The pregnancy was otherwise unremarkable, and the 20-week and 28-week prenatal ultrasound scans detected no abnormalities. The lack of ultrasonographic findings could be attributed to post-traumatic haemorrhage or rapid growth of congenital infantile fibrosarcoma (CIFS) occurring after the last scan.[4]

On examination, a large, left-sided orbital mass with severe proptosis and dystopia was found (*Figure 1*). The mass

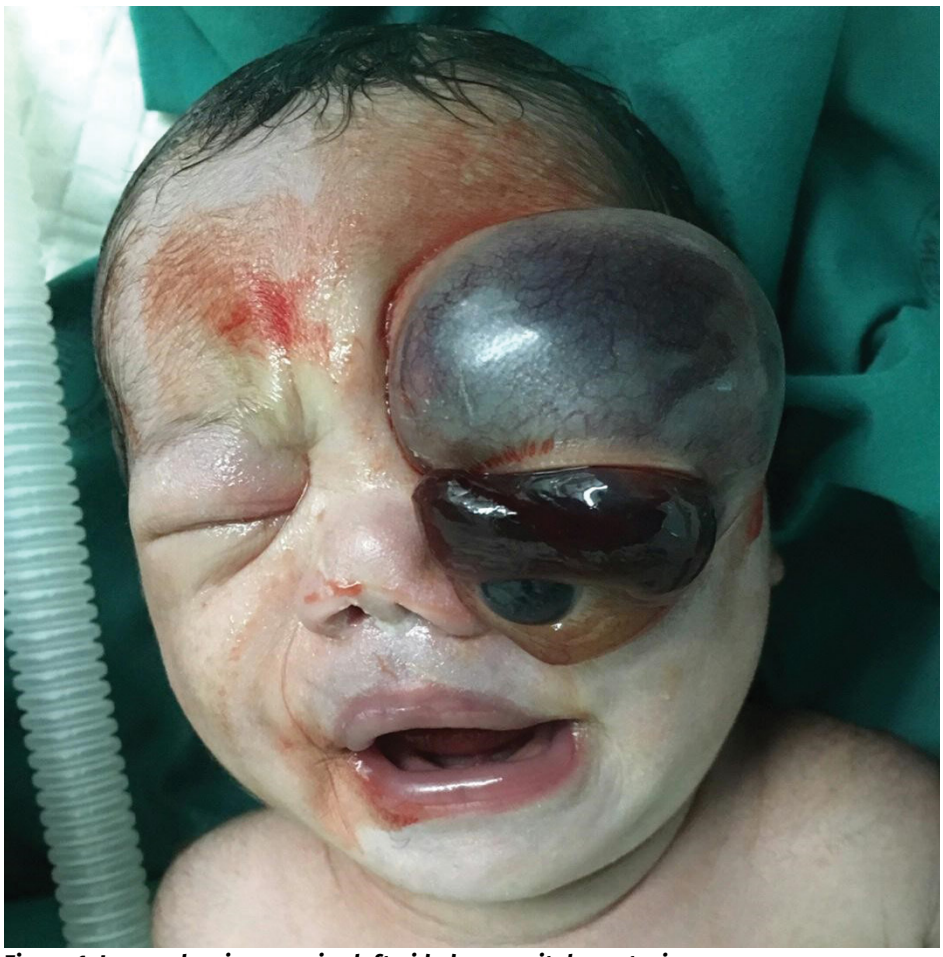

Figure 1. ***Image showing massive left-sided congenital proptosis***

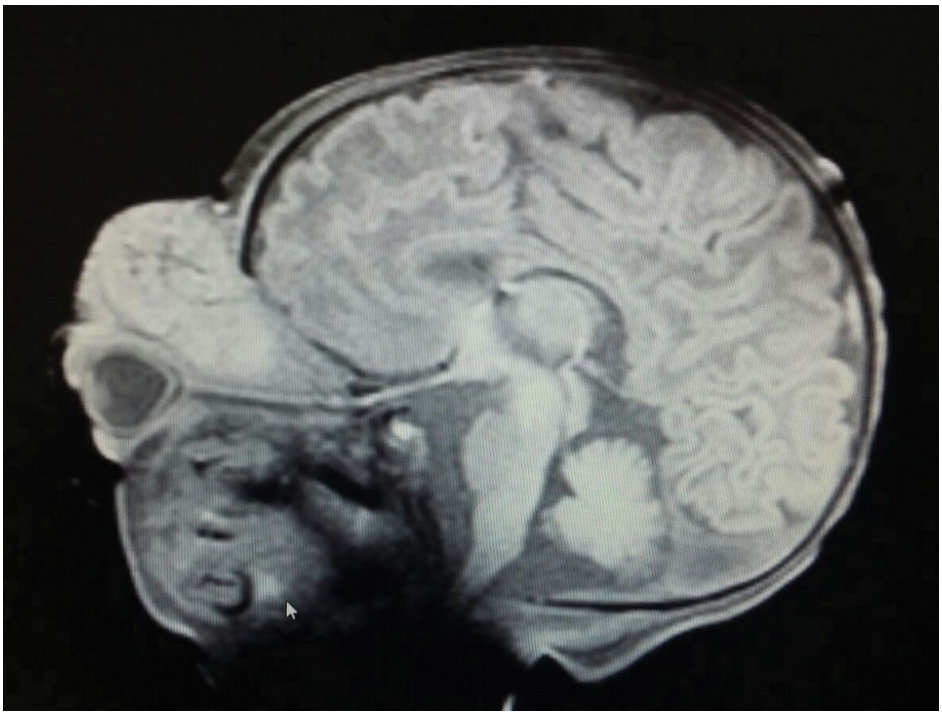

Figure 2. ***MRI image showing an intra-orbital mass causing proptosis and stretching of the optic nerve***

was firm and fixed, had a vascular appearance, was non-pulsatile with no bruit, and was actively oozing a combination of fresh and lysed blood. The differential diagnosis included: traumatic orbital haematoma, malignancy with or without intratumoral haemorrhage, arteriovenous malformation and encephalocoele.

Magnetic resonance angiography showed a large, homogenous, minimally vascular mass filling the left orbital cavity significantly stretching the optic nerve (*Figure 2*). There was no intracranial extension and no bony erosion. Incisional biopsy was performed, and histopathological examination revealed poorly differentiated, spindle-shaped fibroblasts with a herringbone architecture.

The immunohistochemistry report found the tumour to be vimentin positive, desmin negative and S100 protein negative. It was noted that the differential diagnoses included congenital–infantile fibrosarcoma, dermatofibrosarcoma protuberans, and synovial sarcoma; however, in view of the clinical presentation and morphological features, a congenital–infantile fibrosarcoma was favoured.

The patient was seen by a multidisciplinary team including paediatric ophthalmologists, neonatologists, radiologists and oncologists. The decision was taken to initiate neo-adjuvant chemotherapy followed by surgical debulking or complete resection of the tumour. Vincristine was administered via a Broviac line weekly for three weeks, followed by actinomycin D biweekly for three weeks. The tumour started to show signs of involution by the second week. The child was re-admitted on week 3 for Broviac line sepsis which resolved on intravenous antibiotics. Unfortunately, the patient was lost to follow-up and was reported to have died of uncertain causes shortly after the last visit.

Discussion

CIFS is a rare mesenchymal neoplasm primarily occurring in children under 5 years of age. It is usually diagnosed within the first year of life, with approximately 40% of lesions present at birth.[5] A significant in utero complication of CIFS is fatal haemorrhaging of the tumour.[6] Although malignant, CIFS rarely causes distant metastases, thus cure through complete surgical resection is possible. CIFS can however spread locally into the surrounding neurovasculature which can make resection difficult and may result in life-threatening complications. The mainstay of management is early surgical resection with or without neo-adjuvant or adjuvant chemotherapy.[7]

Congenital proptosis may be caused by life-threatening conditions. Early prenatal detection improves outcomes, but its influence on survival has not been studied.[6] One diagnostic modality that has been shown to be effective in detecting such cases is antenatal ultrasound, especially when using high-resolution devices.[2] Early detection allows multi-disciplinary team members to prepare adequately for problems before or at birth.[3] Unfortunately, the proptosis was not detected antepartum in this case. It is not apparent whether this is because the pathology was missed during the prenatal ultrasound scans or whether the pathology only arose after the last ultrasound scan at 28 weeks gestation.

Primary CIFS of the orbit presenting with congenital proptosis is extremely rare. In our literature search, we could only find one other previously reported case.[8]

Ethics statement

The parents of the patient gave informed consent for the case to be published.

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