CASE REPORT

Acting on macrocephaly in the neonatal period: an illustrative case of congenital teratoma

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SUMMARY

An 11-week-old baby was brought to the paediatric emergency department by his mother with a 2-day history of inconsolable crying. On examination, clinical features of macrocephaly, separated sutures and 'sunsetting' of the eyes were noted. Abnormal head circumference measurements had been obtained on several occasions since birth, but were not acted on contrary to local guidance. During the emergency admission, an urgent CT scan revealed a large posterior fossa tumour consistent with a teratoma causing severe obstructive hydrocephalus. Following referral to a neurosurgical centre, emergency ventricular drainage and debulking surgery were performed, unfortunately with no option for cure. Distress to mother and baby could have been reduced with a more timely diagnosis.

GP surgery as her baby seemed intermittently unsettled and the practice nurse made an outpatient hospital referral having noted the enlarged head.

INVESTIGATIONS

Bloods (full blood count, renal profile and clotting profile) taken at presentation to the emergency department were normal. An emergency CT scan (figure 2) revealed a large heterogeneous posterior fossa mass containing fat, solid-cystic elements and calcification with associated severe obstructive hydrocephalus, although no conclusive radiological diagnosis was offered at the time. A subsequent MRI following referral to a tertiary neurosurgical unit confirmed the posterior fossa mass with imaging features of a teratoma.

BACKGROUND

Head circumference measurements are performed as part of neonatal and early infancy routine checks and are a useful tool in screening for macrocephaly.¹ Local and national protocols have been implemented as guidance for appropriate investigation, but these are not always followed. This case illustrates how failure to act on cranial enlargement may lead to a missed tumour diagnosis. Through this example, we hope to raise awareness and improve practice.

CASE PRESENTATION

An 11-week-old baby was brought to the paediatric emergency department by his mother with a 2-day history of inconsolable crying. He had been born at term by emergency caesarean section for fetal bradycardia following an uneventful pregnancy. His mother's booking bloods and antenatal ultrasound scans had been normal.

On examination he was irritable with obvious macrocephaly, separated cranial sutures and 'sunsetting' of the eyes. His head circumference at presentation measured 48.5 cm, which was well above the 99.6th centile (figure 1). There was no history of vomiting or any other preceding illness.

At birth, the baby's head circumference had been measured at 39 cm (just above the 99.6th centile with birth weight on the 25th centile), which prompted a review by the paediatric junior doctor. The head enlargement was attributed to caput succedaneum and the baby was discharged. At the 6-week baby check, the general practitioner (GP) measured the head circumference at 43 cm, but no further action was taken. Two days prior to the acute hospital presentation, the mother visited her

DIFFERENTIAL DIAGNOSIS

The differential diagnosis for macrocephaly is wide including neoplastic and non-neoplastic causes. The rapidly enlarging head circumference in this case raises concern for obstructive hydrocephalus, possible causes of which include a tumour, but also intracranial haemorrhage, infection or an underlying non-tumoural structural lesion such as aqueductal stenosis.

Among the differential for congenital brain tumours are teratoma, primitive neuroectodermal tumour (including medulloblastoma), atypical teratoid rhabdoid tumour, ependymoma, astrocytoma and choroid plexus papilloma. Radiologically, the presence of fat, soft tissue elements and calcification within a lesion is pathognomonic for teratoma.

TREATMENT

Following transfer to the neurosurgical unit, the patient underwent emergency ventricular drainage and posterior fossa debulking surgery. Teratoma was histologically confirmed.

OUTCOME AND FOLLOW-UP

Owing to the large size and location of the tumour, curative resection was unfortunately not an option. Since the initial admission, the patient had undergone repeated surgery to drain cystic components of the tumour and remains alive at age 18 months, albeit with a significant developmental impairment. This length of survival is unusual and could represent an atypical, less aggressive type of teratoma, although there is no histopathological evidence to support this hypothesis. The patient remains under neurosurgical care for palliation.

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Figure 1 Growth chart demonstrating increasingly abnormal head circumference measurements since birth.

DISCUSSION

Congenital brain tumours are uncommon with a prevalence between 1.7 and 13.5/100 000 live-births.² These lesions only represent 0.5–1.5% of all paediatric brain tumours, whereby the supratentorial compartment is affected more commonly than the posterior fossa.^{3 4} Teratoma represents the most frequently encountered congenital brain tumour and has a poor prognosis

with death usually occurring not long after birth.^{5 6} A single recent case report describes the cure of a patient with resection following neoadjuvant chemotherapy.² To the best of our knowledge, there are no other case reports of congenital teratoma presenting in the postnatal period with progressive macrocephaly. However, cranial enlargement is a well-known feature of congenital brain neoplasms and hydrocephalus.¹

Figure 2 Axial CT, axial and sagittal MRI demonstrating the large posterior fossa tumour containing fat and calcification (white arrow) as well as solid-cystic elements (black arrows). There is severe hydrocephalus (H) of the lateral ventricles and third ventricle with displacement of the cerebellum (C) by the tumour.

Diagnosis is important antenatally to prevent obstructed labour and to reduce psychological morbidity in the mother. Unfortunately, the tumour may not always be evident on a 20-week antenatal anomaly ultrasound scan.⁷ Where surgical intervention and/or adjuvant therapy are options, these are most likely to have the maximum impact if an early diagnosis is made. With such a poor prognosis, early planning of palliative care may reduce distress to the child and family.

There were worrying clinical features in this case from birth as well as several missed opportunities for intervention. The mismatch in birth weight (25th centile) and head circumference (above 99.6th centile) should have prompted a senior paediatrician review as per local protocol, but this was not performed. A cranial ultrasound scan examination would have been mandatory

if the clinical examination showed diastatic cranial sutures. At the 6-week check, a clearly abnormal head circumference was documented, but not acted on, and even when the patient returned thereafter with symptoms of decompensating hydrocephalus, only an outpatient referral was made.

This case highlights the importance of acting on neonatal cranial enlargement, which is a requirement as per the UK national screening standard and according to NICE guidelines.^{8,9}

Contributors All the authors equally contributed to the article and have had the opportunity to review the manuscript.

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Learning points

- ▶ Head circumference measurement (OFC) as part of any routine baby check is important, as deviations may be the first indicator of a serious underlying condition including tumour.
- ▶ An abnormal measurement must not only be documented, but always requires further investigation.
- ▶ A large OFC (more than 38 cm) in the presence of full fontanelles or separated sutures on examination must prompt urgent imaging, usually with cranial ultrasound in the first instance.
- ▶ Radiologically, the presence of fat, calcification and soft tissue elements within a tumour is pathognomonic for teratoma meaning a conclusive diagnosis would have been possible on CT scan.

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