Unusual association of diseases/symptoms

Sotos syndrome, failure to thrive and parotitis

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Correspondence to Dr Megan R Thomas, Dr.Thomas@bfwhospitals.nhs.uk**Summary**

Sotos syndrome is characterised by excessive pre and postnatal growth, a variable degree of learning difficulties and a recognisable facial appearance. This report highlights the difficulty in making the diagnosis where failure to thrive is the presenting feature and documents a previously undescribed association with recurrent parotitis.

BACKGROUND

Sotos syndrome was first described in 1964,¹ and since then there have been many publications further delineating the spectrum of difficulties and characteristic features. Mutations or microdeletions of the *NSD1* gene have now been identified in the majority of clinically identified individuals tested. There is a characteristic facial gestalt with a prominent forehead, macrocephaly, sparse hair particularly frontally, downslanting palpebral fissures and a prominent lower jaw often with a pointed chin. Excessive growth is described as having a prenatal onset with length being most strikingly affected while birth weight can be within the normal range.² Occipital frontal circumference is often increased at birth.

Cardiac and genitourinary anomalies, neonatal jaundice, neonatal hypotonia and scoliosis are all commonly found. The scoliosis can present early and progress without treatment. Developmental delay is usually present with delay in gross-motor skills and expressive language being most common. Development often improves over time but most individuals will have some learning difficulties and require additional educational support. It is therefore important to establish the correct diagnosis in order to look for associated conditions and ensure appropriate interventions and support are achieved.

This case highlights an additional differential diagnosis to be considered in children presenting with feeding difficulties, hypotonia and jaundice in the neonatal period. Normal birth weight does not rule out the diagnosis and feeding difficulties can initially lead to marked failure to thrive. Regular growth measurements, awareness of the characteristic facial gestalt and the availability of a gene test should enable the diagnosis to be achieved in a timely manner. It also describes, for the first time, an association with recurrent parotitis.

CASE PRESENTATION

Delivery was at term+6 days by an emergency Caesarean section due to failure to progress following induction of labour because of poor fetal movements and raised maternal blood pressure. Birth weight was 3.04 kg, just above the 9th centile and head circumference 34.5 cm, on the 25th centile on the standard girls growth chart – Child

Health Growth Foundation 1996. It was documented that the skull sutures were overriding and there was marked caput succedaneum. The measured head circumference may therefore be an underestimate. In accordance with local practice length was not measured at birth.

Although the baby initially went to the postnatal ward she was transferred to the special care baby unit at 11 h of age due to failure to establish feeding and a low blood sugar. She was noted to be moderately floppy and to have a systolic murmur. Tube feeds were required for 5 days and moderate jaundice developed on day 2 but never required treatment. She was fully bottle fed by day 6 and discharged home on day 8.

The following evening she was readmitted through Accident and Emergency with poor feeding and mild dehydration. Weight on admission was 2.84 kg, on the 2nd centile. About 36 h after admission, a parotid swelling was noted. No pus was detected on examination. A diagnosis of parotitis was made and she was discharged well after 5 days treatment.

Re-admission occurred on day 24 with a tender, warm, diffuse parotid swelling. Again no pus was noted on examination. Diagnosis was again parotitis and she was discharged home after 4 days treatment to complete the course of antibiotics orally at home.

Concern then concentrated on development, and she was referred to the Child Development Centre at 3 months of age. Smiling was not noted until she was 4 months old and there were initial concerns around vision which resolved. Gross-motor skills were delayed and movements were rather jerky. She had plagiocephaly and habitually held, her head turned to the right. The sternomastoid was noted to be tight though no frank tumour was identified. At 19 weeks of age her weight was between the 0.4th and 2nd centiles, with length and head circumference on the 50th (see figure 1).

Due to concerns about increasing head size, marked frontal bossing and plagiocephaly she was referred for a neurosurgical opinion at a tertiary centre at 9 months of age by the hospital paediatrician. The neurosurgical team elected to review in 6 months.

Growth and development continued to be monitored and at 12 months of age her weight had increased to between the 50th and 75th centiles, while her length and head

Figure 1 Growth chart.

circumference were on the 98th centile. Her forehead was noted to be prominent and her hair generally sparse particularly frontally. A murmur was heard and subsequently confirmed as a small patent ductus arteriosus. On review at 18 months, height and head circumference continued just above the 98th centile with weight lying between the 75th and 91st centile. A presumptive diagnosis of Sotos

syndrome was made, which was subsequently confirmed with gene testing.

INVESTIGATIONS

Investigations performed on the first admission with poor feeding revealed a markedly raised C reactive protein. On the second admission the C reactive protein was again

elevated and the white cell count was $38.6 \times 10^9/l$ with 80% neutrophils. A throat swab produced a profuse growth of *Staphylococcus aureus*.

DIFFERENTIAL DIAGNOSIS

The differential diagnosis for neonatal feeding difficulties, hypotonia and jaundice are multiple including neurological, metabolic and syndromal aetiologies.

TREATMENT

Both episodes of parotitis were treated with intravenous antibiotics.

Intervention for the developmental delay focused on physiotherapy, occupational therapy and speech and language therapy.

The patent ductus arteriosus required surgical ligation.

OUTCOME AND FOLLOW-UP

Due to the diagnosis of Sotos syndrome, the child was discharged from subsequent neurosurgical follow-up as her head shape and increasing head size could be attributed to this condition. Close monitoring for associated complications detected scoliosis at an early stage and treatment is currently with bracing. Early notification of special educational needs has ensured that additional educational support is provided in school.

DISCUSSION

Neonatal feeding difficulties are described in the literature about Sotos syndrome and occurred in 21% of cases in one study,³ and 35% in another.² Failure to thrive is not well documented. However, Cole, in his review of overgrowth syndromes commented that failure to thrive could 'mask' the diagnosis.⁴ The onset of excessive growth is not always prenatal and all growth parameters can fall well within the normal range at birth.

Recurrent parotitis is a rare condition and its aetiology is still not fully understood. It appears to have an association with dehydration and the mechanism postulated is that

the reduced saliva flow allows organisms to track up into the salivary gland.⁵ *S aureus* is the most common organism implicated and generally has a good prognosis. In contrast with suppurative parotitis, where the presence of pus is pathognomonic, pus is typically absent in recurrent parotitis and the role of antibiotics is less clear. The significant, persisting feeding difficulties in this child associated with clinically noted mild dehydration is the likely cause for the two episodes of parotitis. A profuse growth of *S aureus* was obtained from a throat swab suggesting this may have been the causative organism. No further episodes of parotitis have occurred.

Learning points

- Sotos syndrome should be considered in all children presenting with feeding difficulties and hypotonia in the neonatal period regardless of birth weight
- Failure to thrive does not rule out an overgrowth syndrome
- Evaluation of a well child with an increasing head circumference should include consideration of their other growth parameters
- Recurrent parotitis may be due to feeding difficulties and dehydration.

Competing interests None.

Patient consent Obtained.

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