Case report

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Silver Russell syndrome: A suspected syndromic child in Sri Lanka presenting with recurrent head-banging associated with force-feeding

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Introduction

Silver Russell syndrome (SRS) is a heterogeneous genetic syndrome with a worldwide incidence of 1 in 30000 to 1 in 100000 (1). It has significant implications such as intrauterine growth restriction, characteristic facies, global developmental delay, intellectual disability and feeding difficulties (1). The diagnosis is based on clinical findings as per the Netchine Harbison score proposed by Azzi et al. in 2015 (2). Though genetic testing confirms the diagnosis, a significant subset of patients may test negative, in which case the diagnosis cannot be excluded (1-3).

We could only find two previous reports of SRS from Sri Lanka. One report had classical clinical features of SRS, and another had the left second metacarpal pseudo epiphysis (6,7). Behavioural dysregulation and feeding problems related to SRS have not been reported in Sri Lanka. SRS is associated with poor appetite, fussiness, slow feeding, and oral-motor dysfunction (8). Further, the syndrome presents with physical and verbal aggressiveness, oppositional behaviour, inattention, and scholastic deficits (9).

We discuss how a child, likely to be having SRS, presenting with intellectual disability, feeding difficulties and recurrent head banging, was diagnosed using the Netchine Harbison score and was managed.

Case report

Master N* was a five-year-old child who had delayed developmental milestones and short stature. He had walked independently at around three years of age and could still not climb stairs. He spoke the first intelligible word at two years of age and cannot express it in sentences at present. Growing up, the worried parents always struggled to feed him, during which he banged his head on any available surface. Over time, the parents felt that he sometimes banged his head for seemingly no reason, such as hearing the noise of a crow or a passing vehicle. These resulted in minor injuries to the scalp that resolved without interventions.

On presenting to the closest tertiary care centre to his home, he was started on risperidone 0.5mg nocte by a general adult psychiatrist three years ago for his behaviour dysregulation. This medication was continued to date with doubtful compliance, and initial improvement later plateaued. The week before the presentation, the head banging worsened, resulting in a large circumferential scalp hematoma. He was brought to the closest provincial general hospital and later transferred to a tertiary care centre with child and adolescent psychiatry expertise. After surgical assessment and treatment, he was referred to the child and adolescent psychiatry unit to explore any psychiatric causes for recurrent head banging.

N* was of small for gestational age and delivered via emergency caesarean section due to lack of progress. He had febrile convulsions in the past. He had been treated with prednisolone, tailing off doses for nephrotic syndrome at three years of age without recurrences. He was the only child to a father who was a manual unskilled worker with an unstable income and a mother who was a housewife.

The physical examination portrayed failure to thrive (weight and height of less than 3SD for his age), limb length discrepancy of 1cm, prominent forehead, triangular facial appearance, wrinkled skin akin to that of an older man and disseminated tinea corporis as shown in Figure 1. He had a circumferential scalp oedema. The eye contact was responsive. He spoke a few intelligible words in a squeaky voice incompatible with his chronological age.

Several differentials were considered, including SRS and Progeria syndrome (3,12,13,14). We also considered startle epilepsy for the head-banging episodes (15).

Despite speech delay and self-harming, the child demonstrated reasonable social communication skills without restricted, repetitive behaviour. N* had significant reading, writing, and calculation deficits and a limited understanding of daily functions in relation to age, which is suggestive of intellectual developmental disorder. Non-verbal intelligence assessed by the TONI-4 test was in the below-average range.

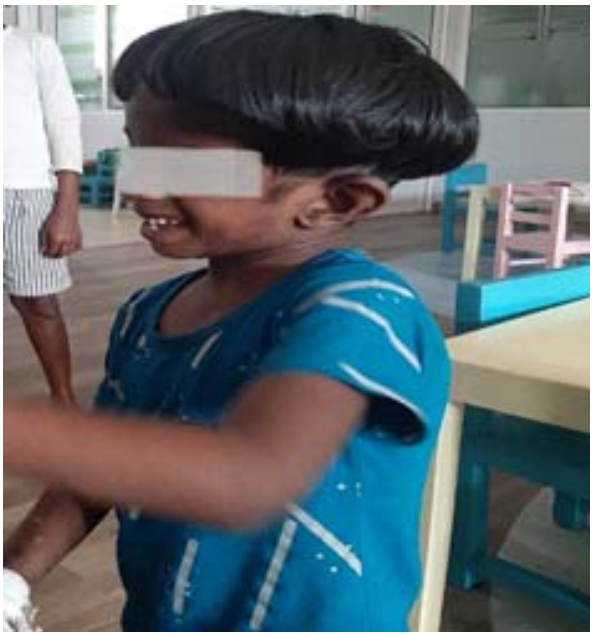

Figure 1. A five-year-old Sri Lankan child suspected to have Silver Russell syndrome.

Paediatric neurology, paediatric endocrinology, clinical genetic, and paediatric dermatology opinions were taken, and an electroencephalogram (EEG) was done. The risperidone dose was up-titrated to 1mg nocte, and an adult psychiatry referral for the mother was taken as she was depressed.

The detailed history showed that the head-banging occurred predominantly when his demands were unmet, when force-feeding, or when he could not get the parents' attention. A frontal epileptogenic focus was there on EEG, as shown in Figure 2, which was not conclusive of epilepsy. The neurology opinion was that the sensory seizures are unlikely and antiepileptics are not required for the child. Silver Russell syndrome was diagnosed according to the Netchine Harbison score, as shown in Table 1 (2).

The clinical geneticist recommended further testing for a definitive diagnosis, which was unavailable in the hospital and not affordable for the parents. On discharge, after evacuating the hematoma, the parents were educated about the genetic diagnosis and to have realistic expectations regarding his development and feeding. Behavioural therapy was initiated to manage the head banging and speech and language therapy for expressive language delay. The adult psychiatrist commenced the mother on sertraline for her depressive disorder.

Table 1. Netchine-Harbison clinical scoring system for Silver Russell syndrome

Clinical criteria	Clinical criteria	Index child
SGA (birth weight and/or birth length)	≤–2 SDS for gestational age	Present
Postnatal growth failure	Height at 24 ± 1 months ≤–2 SDS or height ≤–2 SDS below mid-parental target height	Present
Relative macrocephaly at birth	Head circumference at birth ≥1.5 SDS above birth weight and/or length SDS	Present
Protruding forehead	Forehead projecting beyond the facial plane on a side view as a toddler (1-3 years)	Present
Body asymmetry	LLD of ≥0.5 cm or arm asymmetry or LLD <0.5 cm with at least two other asymmetrical body parts (one non-face)	Present
Feeding difficulties and/or low BMI	BMI ≤–2 SDS at 24 months or current use of a feeding tube or cyproheptadine for appetite stimulation	Present

LLD – leg length discrepancy; SDS – SD score; SGA – small for gestational age

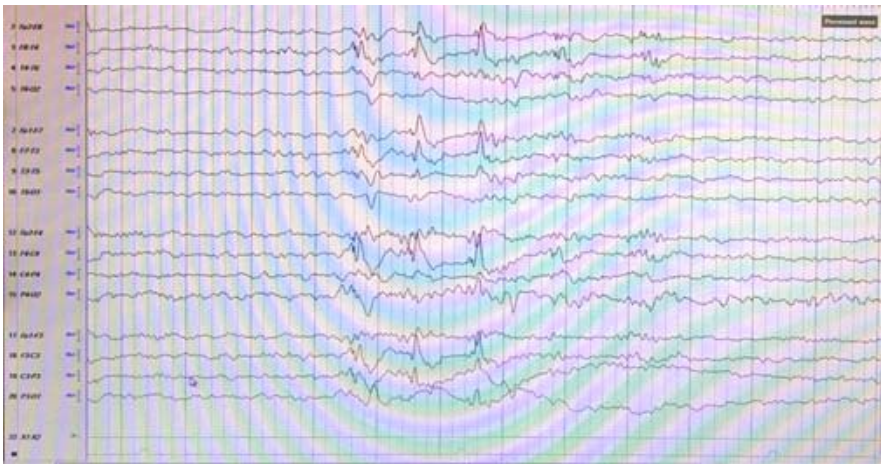

Figure 2. Electroencephalogram of the child with Silver Russell syndrome.

Discussion

Children with Silver Russell syndrome can be fussy feeders for many reasons (11). The parents should be educated to have realistic feeding and achievable growth goals. Force-feeding needs to be avoided due to the risk of metabolic syndrome. Growth hormone therapy is beneficial in some patients (4). The paediatric clinical setup needs to know how to diagnose this rare syndrome clinically. In this child, the poverty of the family, maternal depression and the absence of facilities in the government for genetic diagnosis were our main challenges.

Consensus-based clinical criteria became a boon to proceed with the multidisciplinary management in this circumstance. In SRS, the most commonly recognised molecular causes are loss of methylation on chromosome 11p15 in half of patients and maternal uniparental disomy for chromosome 7 in a minority (1). Even after genetic testing, the underlying molecular cause remains unknown in a third of patients meeting the clinical criteria.

In SRS, there are several clinical problems, such as severe feeding difficulties, reduced postnatal growth, behaviour dysregulation, gastrointestinal symptoms, episodes of hypoglycaemia, issues related to body asymmetry, scoliosis, scholastic deficits and speech delay (9,16). Regional healthcare facilities in Sri Lanka may be unable to address many of these challenging presentations. Therefore, children with SRS could be recognised early using the clinical criteria and referred to tertiary care management.

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None.

Declaration of interests

The authors declare that there is no financial or non-financial conflict of interest.

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