CASE REPORT

Status epilepticus in a child with Sanjad Sakati syndrome

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SUMMARY

We report a 6-year-old boy who presented with status epilepticus, who had facial dysmorphism, growth and mental retardation. On investigation, he had hypocalcaemia, hypoparathyroidism and bilateral calcification of basal ganglia in cranial tomographs; features consistent with Sanjad Sakati syndrome. He was treated with intravenous calcium gluconate initially followed by oral calcium and calcitriol and recovered completely.

BACKGROUND

Sanjad Sakati syndrome (SSS), an autosomal recessive disorder is characterised by hypoparathyroidism, mental retardation, growth retardation and dysmorphic facial features. It is a rare cause of hypoparathyroidism and hypocalcaemia, which leads to status epilepticus in children.¹ The usual causes of hypoparathyroidism are aplasia or hypoplasia of parathyroid gland (Di-George/velocardio-facial defect), x-linked recessive and autosomal dominant hypoparathyroidism, Kearns-Sayre syndrome and mucocutaneous candidiasis. It is mostly reported from Middle East countries.² We report here a case, who had presented to us with status epilepticus, dysmorphic facies and delayed neuro-developmental milestones owing to SSS, which is not reported from India.

CASE PRESENTATION

A 6-year-old boy, born from consanguineous parents, was brought in emergency with a history of fever and multiple episodes of generalised tonic clonic seizures for 2 months and unconsciousness for 4 days. At admission, the child was unconscious (Glasgow Coma Score : 4/15) and had generalised tonic-clonic seizure. In emergency, after securing airways, intravenous lorazepam was given followed by intravenous phenytoin (loading 20 mg/kg followed by 5 mg/kg twice a day) to control seizure. In the past, the child had a similar episode 1 year back and was treated by antituberculous drug (2HRZE 10HR) for tuberculous meningitis. The child had global developmental delay (Neurodevelopmental score: motor and adaptive <20, language 60, personal social 40). Anthropometry showed weight 12.4 kg (expected 20.8 kg) and height 100 cm (expected 115.7 cm), both less than the third percentile. His mother was also sputum-positive pulmonary tuberculosis.

On examination, vitals of the child were stable and had dysmorphic facies (prominent forehead, deep-seated eyes, beaked nose, long philtrum, thin lips, poor dentition) (figure 1). Central nervous

system examination (CNS) revealed increased tone in all limbs with exaggerated deep tendon reflexes and bilateral up-going plantars. Other systemic examination was normal.

INVESTIGATIONS

His complete blood count was normal. ABG showed Ph:7.24, PaO₂: 62 mm Hg, PaCO₂: 28 mm Hg, HCO₃:14 meq/l and ionised calcium: 0.69 mol/l. Cerebrospinal fluid examination showed no organism on gram's and acid-fast bacilli staining; cells 5/mm³ (lymphocyte only); glucose 86 mg/Ddl (blood glucose 108 mg/dl) and protein 36 mg/dl. Cerebrospinal fluid and blood culture for aerobic bacteria were sterile. CT of the cranium showed bilateral basal ganglia calcification (figure 2). MRI of the cranium showed mild cerebral atrophic changes with prominent ventricular system and B/L calcification of basal ganglia (figures 3 and 4). Serum calcium and phosphate values were 5.8 and 6.22 mg/dl, respectively. Serum magnesium concentration was 1.48 mEq/l (normal 1.2–1.9 mEq/l). Based on the report of the investigations, hypoparathyroidism as cause of seizure was suspected and confirmed by serum parathyroid level, which was <3 pg/dl (normal 25–75 pg/dl).

DIFFERENTIAL DIAGNOSIS

Based on the history, possibility of tubercular meningitis was kept but, in the present case, cerebrospinal fluid examination was within normal limit, so tuberculous meningitis was ruled out. The other differential diagnosis is Kenny-Caffey syndrome type 1. In

Figure 1 Facial dysmorphism of affected child.

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Figure 2 Cranial tomograph showing bilateral calcification of basal ganglia.

contrast to SSS, children with Kenny-Caffey syndrome have normal intelligence, delayed closure of anterior fontannele, macrocephaly, postnatal growth retardation, anophthalmos and corneal opacity, which were absent in this case.

TREATMENT

The child was treated with intravenous calcium initially for 72 h, thereafter oral calcium. His consciousness and seizures were controlled with intravenous calcium gluconate. Subsequently, the child was shifted to oral calcium and calcitriol and was discharged on the same.

Figure 3 MRI of the cranium showing prominent ventricular system and B/L calcification of basal ganglia in T1-weighted and T2-weighted images, respectively.

Figure 4 MRI of the cranium showing prominent ventricular system and B/L calcification of basal ganglia in T1-weighted and T2-weighted images, respectively.

OUTCOME AND FOLLOW-UP

The patient had no seizure in follow-up at 1, 3 and 6 months and is attending his school but had facial dysmorphism, global developmental delay and mental retardation.

DISCUSSION

SSS is an autosomal recessive disorder, which was first reported from the Kingdom of Saudi Arabia by Sanjad *et al* in 1988.³ Three years later, its inheritance and chromosomal configuration were confirmed by the same team in King Faisal Specialist Hospital and Research Center, Saudi Arabia.⁴ Till now, cases have been reported from all Arabian countries. No case report, to the best of our knowledge, has been reported from India. Parvari *et al*⁵ reported a gene location on chromosome 1q42–43, caused by mutation of the TBCE gene. Some consider it a variant of Kenny-Caffey syndrome type 1, as both share the same allele.⁶

SSS presents with features of hypoparathyroidism, microcephaly, seizures, IUGR, mild-to moderate mental retardation and specific dysmorphic facies.³⁴ Facial dysmorphism in SSS includes beaked nose, deep-set eyes, depressed nasal bridge, long philtrum, microcephaly, micrognathia/retrognathia, thin lips, large floppy ears and teeth anomalies, as in our case. Seizures of any type, mild mental retardation, short stature/dwarfism, small foot, small hands and cellular immune deficit may be present.⁷ Our case had facial dysmorphism, growth and mental retardation and hypothyroidism. This patient had all the features of SSS, but the facility of mutational analysis for TBCE gene is not available at our institute.

Learning points

- ▶ Status epilepticus is a common paediatric emergency.
- ▶ Sanjad Sakati syndrome (SSS) presenting as status epilepticus is extremely rare.
- ▶ SSS is a rare cause of primary hypoparathyroidism.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

REFERENCES

- 1 Koller J, Zarroung AE, van Heerden J, *et al.* Primary hyperparathyroidism in pediatric patient. *Pediatrics* 2005;115:974–80.
- 2 HersHKovitz E, Shalitin S, Levy J, *et al.* The new syndrome of congenital hypoparathyroidism associated with dysmorphism, growth retardation, and developmental delay: a report of six patients. *Is J Med Sci* 1995;31:293–7.
- 3 Sanjad S, Sakati N, Abu-Osba Y. Congenital hypoparathyroidism with dysmorphic features: a new syndrome. *Pediatr Res* 1988;23:271A.
- 4 Sanjad SA, Sakati NA, Abu-Osba YK, *et al.* A new syndrome of congenital hypoparathyroidism, seizure, growth failure and dysmorphic features. *Arch Dis Child* 1991;66:193–6.
- 5 Parvari R, HersHKovitz E, Grossman N, *et al.* Mutation of TBCE causes hypoparathyroidism-retardation-dysmorphism and autosomal recessive Kenny-Caffey syndrome. *Nat Genet* 2002;32:448–52.
- 6 Naguib KK, Gouda SA, Elshafey A, *et al.* Sanjad–Sakati syndrome/Kenny–Caffey syndrome type 1: a study of 21 cases in Kuwait. *East Mediterr Health J* 2009;15:345–52.
- 7 Kelly TE, Blanton S, Saif R, *et al.* Confirmation of the assignment of the Sanjad–Sakati (congenital hypoparathyroidism) syndrome (OMIM 241410) locus to The dysmorphic facies of the second case, chromosome 1q42–43. *J Med Genet* 2000;37:63–4.

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