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Case report

Sanjad-Sakati syndrome with corneal opacity in a Palestinian neonate: case report

Allam Fayez Abuhamda[1], Aymen Mohammed Elsous[2,3]

[1]NICU Department, Shifa Hospital, Gaza Strip, Palestine
[2]Faculty of Health Professions, Israa University, Gaza Strip, Palestine
[3]Unit of Planning and Policy Formulation, Ministry of Health, Gaza Strip, Palestine

Abstract

Sanjad-Sakati syndrome is an autosomal recessive disorder found mainly in people of Arabian origin. This is a report of a Palestinian premature (35 weeks gestation) newborn who was part of twins and had this rare disease. The syndrome comprises congenital hypoparathyroidism, hypocalcaemia, seizure, severe growth retardation, low IQ and typical facial features. In addition, the baby had corneal opacity, although his elder brother with the same syndrome had not. Supportive treatment in the form of oral vitamin D and calcium are often offered to these children to treat hypocalcaemia.

Keywords

Sanjad-Sakati syndrome, corneal opacity, Palestinian premature newborn, hypoparathyroidism.

Corresponding author

Allam Fayez Abuhamda, Consultant Neonatologist, NICU Department, Shifa Hospital, Gaza Strip, Palestine; mob.: +970 567860508, +970 597502720; email: allam570@yahoo.com.

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Introduction

Sanjad-Sakati syndrome is well known in the Middle East and the Arabian Gulf countries [1-7]. It results from mutations of the gene encoding tubulin-specific chaperone E (TBCE gene) located on chromosome 1q42-q43. Sanjad-Sakati syndrome is characterized by intrauterine growth restriction, hypoparathyroidism, hypocalcemia, hyperphosphatemia, seizure, mental retardation, failure to thrive, long face, depressed nasal bridge, peaked nose, small eyes, small hands, micrognathia and floppy ears. It is associated with high mortality and morbidity resulted from seizure-related hypocalcaemia. It is interesting to find out two siblings with the same genetic mutation but with different eye findings, as the second sibling had corneal opacity while the first one had not. It is dramatic to see more than one sibling with the same genetic disease. Early diagnosis is important not only to treat the case but also to prevent recurrence by genetic testing and family counseling.

Case report

The parents were cousins, had two healthy sons and another six years old male suffered from global developmental delay and characteristic feature of Sanjad-Sakati syndrome confirmed by genetic study (**Fig. 1**).

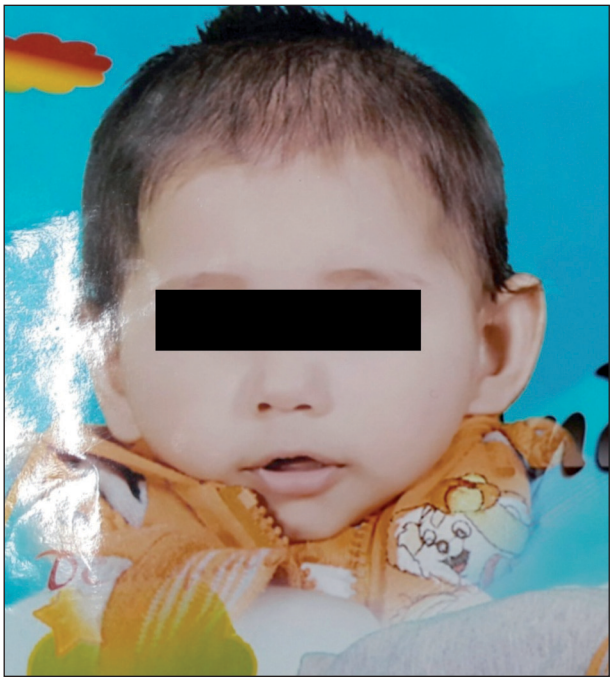

Figure 1. The first sibling with Sanjad-Sakati syndrome, without corneal opacity. Age: 5.5 years old.

The second syndromic sibling was part of dizygotic twins. At the 17th weeks of gestational age, ultrasound showed a fetus smaller than the other one (**Fig. 2**); amniocentesis and genetic study showed that the patient was homozygous for the mutation (c.155-166del12;p.del52-55) in the TBCE gene, responsible for Sanjad-Sakati syndrome.

The twins were delivered at 35 weeks of gestational age via normal vaginal delivery. The first twin was female and had a normal physical examination. The second twin was male; birth weight was 1,300 grams (below the 3rd percentile), head circumference was 29 cm (just above the 3rd percentile), Apgar scores at 1 and 5 minutes were 7 and 9, respectively. He had dysmorphic features; long face, microphthalmia, corneal opacity (**Fig. 3**), depressed nose bridge, peeked nose, long philtrum, thin lips, micrognathia, small hands, small feet, normal male genitalia and large floppy ears (**Fig. 4**).

At the age of 6 days, blood investigation showed serum calcium 6.7 mg/dl (normal range 8-11), serum phosphorus 8 mg/dl (normal range 2.5-5), parathyroid hormone 0.4 pg/ml (normal range 15-68.3); blood sugar was normal. Complete blood count: WBC 25.8 K/UL (normal range 4.1-10.9), HGB 15.8 g/dl (normal range 12-18), RBC 4.5 M/ul (normal range 4.2-6.3), PLT 209 k/ul (normal range 150-450).

The baby received intravenous calcium gluconate to treat hypocalcemia, and when serum calcium became normal oral maintenance of calcium and one alpha Alfacalcidol drops were

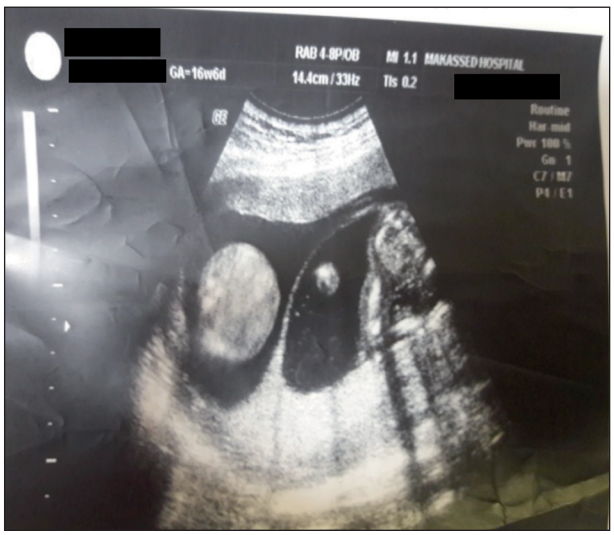

Figure 2. Antenatal u/s at 16 weeks gestation showed asymmetrical twins.

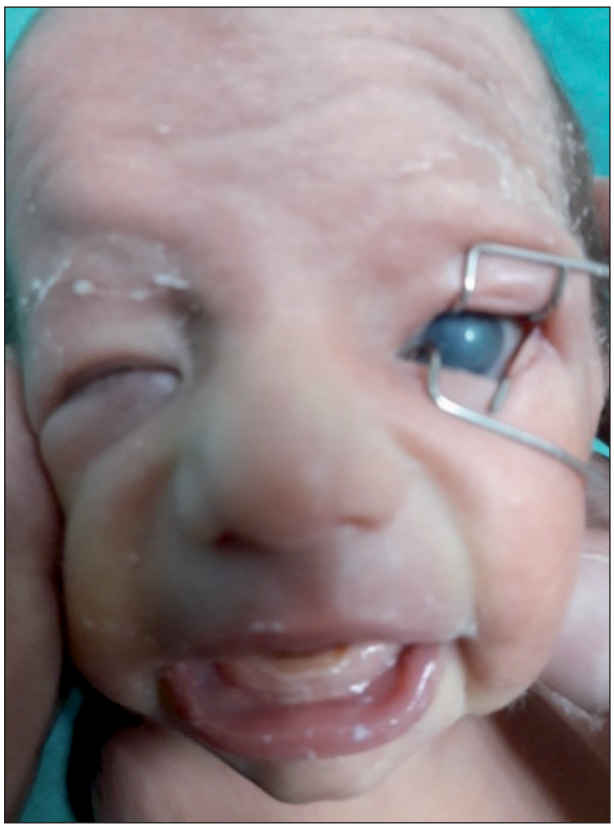

Figure 3. Left eye corneal opacity.

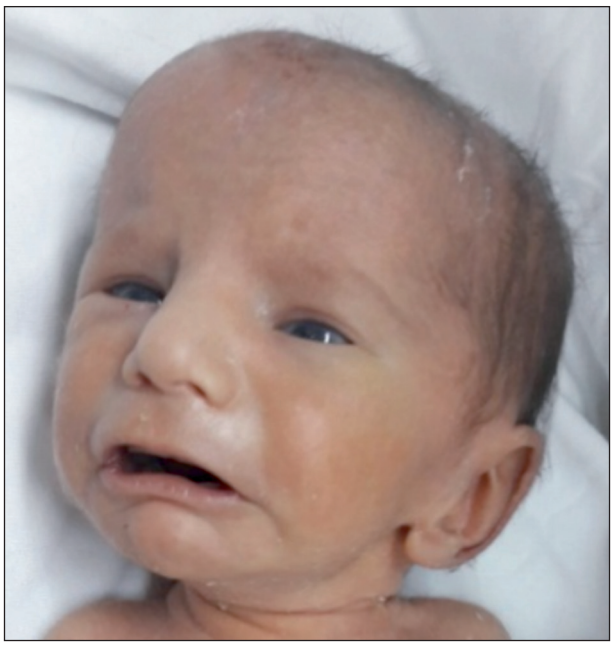

Figure 4. Typical dysmorphic facial feature of Sanjad-Sakati syndrome.

initiated. The baby was discharged home in good general condition at the age of 15 days. Paediatric endocrinologist and ophthalmologist follow-ups were recommended.

Brain ultrasonography, abdominal ultrasonography, echocardiography and skeletal survey X-ray were normal.

The baby passed away at the age of 4 months in the Paediatric Intensive Care Unit due to septic shock.

Discussion

Sanjad-Sakati syndrome is a very rare autosomal recessive congenital disorder with equal distribution in both sexes. The syndrome is linked to chromosome 1q42-q43 and caused by mutations in the TBCE gene. Indeed, with regard to congenital hypoparathyroidism, there is a clinical overlapping between Sanjad-Sakati syndrome and other congenital disorders such as Kenny-Caffey syndrome type 1 and 2 (KCS1 and 2) [8], DiGeorge syndrome and familial isolated hypoparathyroidism. In particular, KCS1 is also caused by the same gene involved in Sanjad-Sakati syndrome, and in this syndrome corneal opacity is present. However, our cases were diagnosed as Sanjad-Sakati syndrome by full clinical criteria, laboratory findings [9], and were confirmed by genetic mutation (c.155-166del12;p.del52-55) in the TBCE gene.

Two Palestinian cases were reported by AbuDraz in the European Hospital in Gaza [10] with a typical feature of the syndrome. Both cases also had no corneal opacity and the genetic study was not done due to limited local resources.

Our case was the first case in Palestine to be diagnosed by physical examination, laboratory investigation and genetic study as a Sanjad-Sakati syndrome with corneal opacity. Although the two siblings had the same gene mutation, they had different eye findings, as the second sibling had corneal opacity since birth and the first one had not. Cases with corneal opacity need follow-up with a paediatric ophthalmologist and fundal examination of both eyes under general anaesthesia to find out if there are more eye abnormalities [11].

Conclusion

Eye findings could be different from one sibling to another with the same gene mutation of the Sanjad-Sakati syndrome. Our patient is the first case of Sanjad-Sakati syndrome with corneal opacity reported in Palestine. The prenatal diagnosis was important for early treatment and prevention of hypocalcaemia. Genetic study is

helpful for family counseling to avoid the same cases in the future, as these patients have high morbidity and mortality.

Declaration of interest

The Authors declare that there is no conflict of interest.

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