Kufa Medical Journal | Vol.18, No.2, 2022

Original article | DOI: https://doi.org/10.36330/kmj.v18i2.3685

Submitted at: 12 June 22
Accepted at: 28 Aug. 22

Sanjad-Sakati Syndrome in Al Najaf Governorate

Mahir Mohammed Ali Altaher [1]

[1] General Pediatrician, Al Zahraa teaching hospital, Al Najaf, Iraq. mahirmmaj@yahoo.com

Abstract

Sanjad-Sakati syndrome is an inherited autosomal recessive disorder that presents primarily in the Arab countries, with its real prevalence not well established. It is a combination of specific facial dysmorphology, failure to thrive, hypoparathyroidism, and variable mental disability which is an unfortunately incurable disease with fatal outcomes .

Keywords: Sanjad-Sakati syndrome, case report, rare diseases.

Background:
Sanjad-Sakati syndrome (SSS), also known as Richardson-Kirk syndrome, has been classified as hypoparathyroidism-retardation-dysmorphism (HRD) syndrome in the Online Mendelian Inheritance of Men (OMIM) no. 241410 [1]. It is an autosomal recessive disorder that was initially described by Sanjad et al. in 1988 [2]. It affects both sexes equally and contains a gene on chromosome 1q42-q43. It has severe and often deadly consequences [4].

This disorder was found almost exclusively in people of Arab origin [5], with reported patients who were Saudi Arabian, Qatari, Palestinian, Kuwaiti, and Omani [6].

Although the exact prevalence is unknown, the estimated incidence in Saudi Arabia ranges from 1:40000 to 1:600000 live births [7]. The syndrome is characterized by dysmorphic features, short stature hypoparathyroidism, and mild to moderate mental retardation [6].

Case presentation

Case 1:
A four-and-a-half-year-old girl from a rural area presented with an attack of loss of bowel motion and vomiting lasting 2 days with refusal to feed. The family reported two previous attacks of acute gastroenteritis and a history of two attacks of seizures from early infancy; each attack lasted a few minutes with no change in the level of consciousness. She was delivered by vaginal delivery with a birth weight of 2 Kg .Regarding the family history, it has been found that the child had one dead sibling with the same clinical features, they reported the cause of death to be pneumonia. The child also has two alive siblings who were the product of consanguineous marriage (second-degree relatives). Besides, there were two nephews in the family with the same clinical features, one of them died at 4 with the cause of death unknown; furthermore, the family reported that she had a delay in walking time, speaking few words only and

she had some difficulties with obeying commands.

On examination, the child looks conscious and, with some dehydration, smaller for her age, with failure to thrive. She also had some odd facial features (small deep-seated eyes, long philtrum, poor dentition, and depressed nasal bridge), and small hands and feet.

Growth parameters: weight 9 kg (Z score -5.8), height 80 cm (Z score -5.7), and head circumference 46 cm (Z score -4). New investigations revealed a white blood cell count of 21100× 109/L (neutrophils 46.2%), hemoglobin 9.9 gm/dl, platelets 310000× 109/L, and C-reactive protein-positive while her previous investigations showed thyroid function test 4.7 µIU/ (0.25-5), T3= 1.19 nmol/l (0.92-233), T4= 96 nmol/l (60-120). Serum calcium is 7 mg/dl and the ionized fraction is 1.8, Parathyroid hormone level is less than 1 Pg/ml (8.7-79.6).

The echocardiogram report was normal and bone age was delayed (less than 1 year). She is on one alpha drop and calcium supplementation.

Due to the typical clinical and laboratory features, she is diagnosed with SSS although genetic testing was not done due to limited resources.

Figure 1: Phenotypes of Sanjad-Sakati syndrome (SSS) in case number one (small deep-seated eyes, long philtrum, poor dentition, and depressed nasal bridge).

Case 2

A nine-month-old male infant presented with repeated vomiting and diarrhea with some dehydration. According to case history, he had a previous attack of seizure in early neonatal life due to low calcium in the blood. His family history reveals that he is the younger brother of the aforementioned case 1. While according to developmental history, he cannot sit yet but he can roll from back to front and the reverse way.

On examination, he is conscious, alert, looks smaller than expected, and his vital signs are within normal for his age and sex. Growth parameter reads: weight 4Kg (Z score=-7), head circumference is 38 cm (Z score=-5.6) and height is 63cm (Z score=-4). He has a small head with deep-seated eyes, a depressed nasal bridge, large ears, and dentition didn't start yet. There are no visible skin lesions and his

chest and abdominal examination were unremarkable His investigations showed that the serum calcium is 7.1 mg/dl, phosphate is 5.5 mg/dl, alkaline phosphatase is of 143 U/l., serum parathyroid hormone level is 2.1 Pg/ml (8.7-79.6), and the echocardiogram was normal. He was put on calcium supplementation and one alpha drop.

Figure 2: Small head with deep-seated eyes, a depressed nasal bridge, and large ears in case number two.

Discussion

Sanjad-Sakati syndrome is a rare autosomal recessive disorder that is described in the Arab world. The mutation is in the tubulin-specific chaperone E (TBCE) gene in chromosomal area 1q42-q43 [5].

This syndrome may share the dysmorphic features with other syndromes namely DiGeorge syndrome and Kenny-Caffey Syndrome [6] although SSS and Kenny-Caffey syndrome have a mutation in the TBCE gene [8], the difference is that patients with Kenny-Caffey Syndrome have normal intelligence, exhibit macrocephaly, and the anterior fontanelle is large and closes late in KCS type 2 patients[9]. On the other hand, DiGeorge syndrome is characterized by hypoparathyroidism and has also T cell immunodeficiency and congenital cardiac anomalies, the two features that are not present in SSS [10].

The facial features of patients with SSS have been shown in many studies and observations of microcephaly, deep-seated eyes, and short stature[2,11] . Errors of refraction, retinal vascular tortuosity, strabismus, and corneal opacities are some of the ophthalmic symptom [2].

There is no cure for SSS but the treatment of its patients is to control the high serum phosphate levels, and the adverse effects of therapy including generalized calcifications. This is done usually by giving calcium supplementation intravenously in case of seizure or orally plus the active form of vitamin D.

Conclusion:

Sanjad-Sakati syndrome is an autosomal recessive disease that is not uncommon in the Arab population. Although there is no

cure for this syndrome, the family can be given genetic counseling and prevent comorbidities. Due to limited resources, genetic testing was not done for these siblings, hopefully, it can be arranged in the future for them.

References:

1. Bashar M, Taimur M, Amreek F, et al. (June 22, 2020) Endocrinological Manifestations of Sanjad-Sakati Syndrome. Cureus 12(6): e8770. DOI 10.7759/cureus.8770

2. Albaramki et al. Sanjad Sakati syndrome: a case series from Jordan. Eastern Mediterranean Health Journal. Vol. 18 No. 5. 2012. P 527-531

3. Osamah Abdullah AlAyed. Sanjad-Sakati Syndrome and Its Association with Superior Mesenteric Artery Syndrome. Hindawi Publishing Corporation Case Reports in Pediatrics Volume 2014, Article ID 108051, 3 pages http://dx.doi.org/10.1155/2014/108051

4. M. Hafez et al. Sanjad Sakati Syndrome: Case reports from Egypt. Egyptian Pediatric Association Gazette 65 (2017) 6–9.

5. Touati et al. Additional Tunisian patients with Sanjad–Sakati syndrome: A review toward a consensus on diagnostic criteria. Archives de Pédiatrie.Volume 26, Issue 2. February 2019, Pages 102-107.

6. Bushra Rafique and Saif Al-Yaarubi. Sanjad-Sakati Syndrome in Omani children. Oman Med J. 2010 Jul; 25(3): 227–229. DOI: 10.5001/omj.2010.63

7. Bashar M, Taimur M, Amreek F, et al. (June 22, 2020) Endocrinological Manifestations of Sanjad-Sakati Syndrome. Cureus 12(6): e8770. DOI 10.7759/cureus.8770

8. AHMAD S TEEBI. Hypoparathyroidism, retarded growth and development, and dysmorphism or Sanjad-Sakati syndrome: an Arab disease reminiscent of Kenny-Caffey syndrome. J Med Genet 2000; 37:145

9. Kenny-Caffey syndrome - NORD (National Organization for Rare Disorders). (2012). Accessed: May 11, 2020: https://rarediseases.org/rare-diseases/kenny.

10. AL-MALIK M.I. The dentofacial features of Sanjad–Sakati syndrome: a case report. International Journal of Paediatric Dentistry 2004; 14: 136–140.

11. Naguib KK, Gouda SA, Elshafey A, Mohammed F, Bastaki L, Azab AS, Alawadi SA. Sanjad–Sakati syndrome/Kenny–Caffey syndrome type 1: a study of 21 cases in Kuwait. Eastern Mediterranean Health Journal, Vol. 15, No. 2, 2009.345-352