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Case Report

Sanjad-Sakati Syndrome in Moroccan Children -

**Mohamed Sellouti*, Ibtissam Belarbi, Salahiddine Saghir, Anas Ayad,
Rachid Abilkassem and Aomar Agadr**

*Faculty of Medicine and Pharmacy, Neonatal Medicine and Intensive Care Unit, Mohammed V
Military Teaching Hospital, Rabat*

***Address for Correspondence:** Mohamed Sellouti, Faculty of Medicine and Pharmacy, Neonatal
Medicine and Intensive Care Unit, Mohammed V Military Teaching Hospital, Rabat, Morocco, Tel :
+212-002-126-606-274-66; E-mail: msellouti@gmail.com

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ABSTRACT

Sanjad-Sakati syndrome is an autosomal recessive that first reported in the Kingdom of Saudi Arabia in 1988. It is characterized by severe growth and mental retardation, congenital hypoparathyroidism associated with seizures and a dysmorphic features. Sanjad-Sakati syndrome is caused by mutations in the gene-encoding Tubulinspecific Chaperone E *TBCE* gene on chromosome 1q42.3. This is a report of a family with this rare disease in Morocco. Supportive treatment in the form of vitamin D and growth hormone supplementation is often offered to patients suffering from Sanjad-Sakati syndrome.

Keywords: Hypoparathyroidism; Facial dysmorphism; Mental retardation

INTRODUCTION

Sanjad Sakati syndrome also known as hypoparathyroidism-retardation-dysmorphism syndrome is an autosomal recessive disorder that was first described in the Kingdom of Saudi Arabia in 1988 [1].

Children affected with this syndrome are characterized by congenital hypoparathyroidism, growth and mental retardation with distinct phenotypic features. Sanjad Sakati syndrome is caused by mutations in the gene-encoding Tubulinspecific Chaperone E (*TBCE*), located on chromosome 1q42.3 [2,3].

They have typical physical features comprised of a long narrow face, deep set small eyes, beaked noses, and large ears that have tendency to droop and undersized jaws (micrognathia). Children suffering from Sanjad Sakati also exhibit mild to moderate mental retardation, leading to poor life prospects [3,4].

CASE 1

A Moroccan boy was presented to the Pediatrics department at Mohamed V military instruction hospital in Rabat at the age of 2 months with generalized tonic clonic convulsions with no history of trauma or fever.

The patient had typical facial dysmorphism, consisting of prominent forehead, deep set eyes, abnormal external ears, microcephaly, microphthalmos, thin upper lip, beaked small nose, micrognathism, and small hands and feet (Figure 1). Systemic examination including the cardiovascular system was normal.

Investigations done at that time revealed Ca: 6.5 mg/dl (normal range: 9-11 mg/dL), PO₄: 8 mg/dl (normal range: 2.4-4.5 mg/dL), ALP: 132 U/L (normal range: 55 U/L-260 IU/L), PTH < 0.4 pmol/L (normal range: 1.2-7.2 pmol/L). Complete blood count, liver functions, renal functions, and urine analysis were within normal limits. The patient was diagnosed as primary hypoparathyroidism and started calcium and vitamin D therapy and convulsions were controlled. Informed consent was obtained from the parents to the genetic study. Genetic analysis of the case revealed mutation: 12 bp (155-166del) deletion within *TBCE* gene in exon 3.

CASE 2

A premature baby boy with gestational age of 35 weeks was born spontaneously vaginal delivery to a G2P2 mother. The mother was seronegative for HIV, Hepatitis B and VDRL. The Apgar scores were 7 and 8 at 1 and 5 minute respectively. The infant weight 2.4 kg (25th-50th percentile), measured 38 cm (50th-75th percentile), and had a head circumference of 30 cm (50th-75th percentile). On clinical exam, the child had facial dysmorphism in the form of microcephaly, deep set eyes, beaked nose, abnormal ear and micrognathia, short hands, and feet (Figure 2). Investigations revealed Ca: 9.4 mg/dl (normal range:



Figure 1: Dysmorphic facies of case 1.



Figure 2: Dysmorphic facies of case 2.

9-11 mg/dL), PO₄: 5.3 mg/dl (normal range: 2.4-4.5 mg/dL), ALP: 59 U/L (normal range: 55 U/L-260 IU/L), PTH < 0.4 pmol/L (normal range: 1.2-7.2 pmol/L). Complete blood count, liver functions, renal functions, and urine analysis were within normal limits.

He was investigated because of family history on the five day of life, She also showed the same biochemical picture: Ca : 6.5 mg/dl, PO₄-4.1 mg/dL and PTH < 0.4 pmol/L. A clinical impression of Sanjad-Sakati syndrome was made due to classical dysmorphism, metabolic derangement, growth retardation and seizures. Baby was put on enteral and parenteral calcium, Vitamin D supplementation. He had one episode of hypocalcemic convulsion, requiring intravenous calcium. He was also started on oral calcium and alphacalcidol. Later on calcium was stopped and she maintained her serum calcium on alphacalcidol only. Genetic analysis of the case revealed mutation: 12 bp (155-166del) deletion within *TBCE* gene in exon 3.



DISCUSSION

Sanjad Sakati Syndrome or hypoparathyroidism-retardation-dysmorphism syndrome is a rare autosomal recessive syndrome and was first described in the Middle East population of Arab origin; reported patients were from Saudi Arabia, Qatar, Israeli Arab, Kuwait, Oman, Morocco and Tunisia [1,3,5,6,7,8]. It is characterized by prenatal and postnatal growth retardation and seizures. Children affected with this syndrome have typical facial dysmorphism, consisting of prominent forehead, deepset eyes, abnormal external ears, microcephaly, microphthalmos, thinned upper lip, hooked small nose, micrognathism, and small hands and feet. Usually they are symptomatic in the new born period with complications of hypocalcaemia and investigations reveal a picture of hypoparathyroidism. These metabolic disturbances are responsible for nephrocalcinosis, medullary stenosis of long bones, other skeletal defects and convulsions. Genetically this disorder has been mapped to the long arm of chromosome 1 (1q42-q43). Mutations in the gene coding for Tubulin Specific Chaperone E (TBCE) have been identified as the cause of the disease in Arabs. However in 2006 Courten et al. [9] described a 4.5-year-old girl with the syndrome who did not have a mutation in the TBCE gene and found another possible gene locus.

Some of the features of this disease resemble Kenny-Caffey Syndrome caused by mutation in the TBCE gene, that shares similar phenotypic features but in addition has osteosclerosis, medullary stenosis of long bones, and normal intelligence

The treatment of patients with Sanjad Sakati syndrome is a challenge for most physicians especially in controlling their high phosphate levels, and the adverse effects of therapy. Early recognition of the disease will lead to proper treatment of patients and prevent associated comorbidities

CONSENT

Written informed consent was obtained from the patient for publication of this case report.

DISCLOSURE

This clinical case was written based on clinical observation without any funding.

REFERENCES

1. Sanjad SA, Sakati NA, Abu-Osba YK, Kaddoura R, Milner RD. A new syndrome of congenital hypoparathyroidism, severe growth failure, and dysmorphic features. *Arch Dis Child.* 1991 Feb;66(2):193-6. doi: 10.1136/adc.66.2.193. PMID: 2001103; PMCID: PMC1792808.
2. Parvari R, Hershkovitz E, Kanis A, Gorodischer R, Shalitin S, Sheffield VC, Carmi R. Homozygosity and linkage-disequilibrium mapping of the syndrome of congenital hypoparathyroidism, growth and mental retardation, and dysmorphism to a 1-cM interval on chromosome 1q42-43. *Am J Hum Genet.* 1998 Jul;63(1):163-9. doi: 10.1086/301915. PMID: 9634513; PMCID: PMC1377236.
3. Kelly TE, Blanton S, Saif R, Sanjad SA, Sakati NA. Confirmation of the assignment of the Sanjad-Sakati (congenital hypoparathyroidism) syndrome (OMIM 241410) locus to chromosome 1q42-43. *J Med Genet.* 2000 Jan;37(1):63-4. doi: 10.1136/jmg.37.1.63. PMID: 10691411; PMCID: PMC1734456.
4. Sanjad SA, Sakati NA, Abu-Osba YK, Kaddoura R, Milner RD. A new syndrome of congenital hypoparathyroidism, severe growth failure, and dysmorphic features. *Arch Dis Child.* 1991 Feb;66(2):193-6. doi: 10.1136/adc.66.2.193. PMID: 2001103; PMCID: PMC1792808..
5. 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. *East Mediterr Health J.* 2009 Mar-Apr;15(2):345-52. PMID: 19554981.
6. Rafique B, Al-Yaarubi S. Sanjad-Sakati Syndrome in Omani children. *Oman Med J.* 2010 Jul;25(3):227-9. doi: 10.5001/omj.2010.63. PMID: 22043344; PMCID: PMC3191633.
7. Richardson RJ, Kirk JM. Short stature, mental retardation, and hypoparathyroidism: a new syndrome. *Arch Dis Child.* 1990 Oct;65(10):1113-7. doi: 10.1136/adc.65.10.1113. PMID: 1701077; PMCID: PMC1792332.
8. Kerkeni E, Sakka R, Sfar S, Bouaziz S, Ghedira N, Ben Ameer K, Ben Hmida H, Chioukh FZ, Ghédira ES, Gribaa M, Monastiri K. Sanjad-Sakati syndrome in a Tunisian child. *Arch Pediatr.* 2015 Sep;22(9):951-5. doi: 10.1016/j.arcped.2015.06.003. Epub 2015 Jul 29. PMID: 26231322.
9. Courtens W, Wuyts W, Poot M, Szuhaï K, Wauters J, Reyniers E, Eleveld M, Diaz G, Nöthen MM, Parvari R. Hypoparathyroidism-retardation-dysmorphism syndrome in a girl: A new variant not caused by a TBCE mutation—clinical report and review. *Am J Med Genet A.* 2006 Mar 15;140(6):611-7. doi: 10.1002/ajmg.a.31122. PMID: 16470743.