Sanjad-Sakati Syndrome in 35-day-old Iraqi Infant: A Case Report

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Abstract

Sanjad-Sakati Syndrome is a very rare autosomal recessive genetic disorder that present solely in Arabian people. This syndrome was discovered for the first time in the kingdom of Saudi Arabia in 1988. Our rare case was reported from the Fallujah Teaching Hospital for Women and Children in Fallujah City, Iraq. This syndrome is associated with hypoparathyroidism, growth retardation, a typical facial appearance, and a variable degree of mental retardation. This prescribed syndrome is usually caused by genetic mutations of tubulin-folding cofactor E (TBCE;604934), which is located on the 1q42.3 chromosome. This case was confirmed by a genetic study which reveals a pathogenic variant in homozygous state in the TBCE gene. Typically, the physical features of this case involve (microcephaly, a long and narrow face, a beaked nose, deep-seated eyes, large ears, thin lips, a long philtrum, micrognathia, small hands and feet). Although Sanjad-Sakati Syndrome is an incurable disease, early diagnosis helps with appropriate palliative treatment of the patient, reducing other associated metabolic and electrolyte disturbances, and allows for a genetic study and counseling of the family, especially in our society due to the high rate of consanguinity.

Introduction

Sanjad-Sakati Syndrome is characterized by hypoparathyroidism, retardation, and dysmorphism; it is an autosomal recessive inheritance that has only been observed in patients of Arabic origin [1]. First, it was reported by Sanjad et al. [2]. Sanjad-Sakati syndrome is typically caused by genetic mutations in the tubulin-folding cofactor E (TBCE; 604934) gene, which is located directly on 1q42.3 chromosome [3].

This disorder is characterized usually by inherent hypoparathyroidism that leads to hypocalcemic seizures of early onset, intrauterine onset of growth delay, craniofacial dysmorphism, and retardation of mentality. The typical physical appearance includes a narrow and long face, a beaked nose, deep-seated eyes, large ears, a thin, long philtrum, and micrognathia [4],[5],[6],[7],[8]. This case was reported from the Fallujah Teaching Hospital for Women and Children in Fallujah City. Our case is a 35-day-old female patient who presented with generalized tonic-clonic seizures but without any history of fever or trauma. Investigations of the patient showed serum calcium of 1 mmol/l (normal range 2.3–2.65 mmol/l), alkaline phosphatase of 199 u/l (normal range 90–180 u/l), phosphorus of 3.05 mmol/l (normal range 1.4–2.5 mmol/l), and parathyroid hormone of 1.20 ng/l (normal range 10–65 ng/l). The complete blood count, renal function, liver function test, and urine examination were all within the normal ranges. Abdominal ultrasound, Echo study were normal. The diagnosis is confirmed by a genetic study through whole exome sequence (WES) by CENTOGENE Company in Germany that revealed TBCE, c.155_166del p. (ser52_Gly55del).

This case was diagnosed as primary hypoparathyroidism and treated with parenteral anticonvulsants, calcium, and vitamin D supplements; seizures were then controlled. She was a term baby, delivered vaginally with a 2 kg birth weight, the fourth offspring of a consanguineous marriage with a history of one abortion.
Length, weight, and head circumference were less than -2 SD from the mean for her age. This patient had typical physical findings consisting of microcephaly, a narrow and long face, a beaked nose, deep-seated eyes, large ears, a long philtrum, a small hand, and small feet. The systemic examination, including the cardiovascular system, was normal.

**Figure 1:** The baby had a beaked nose, large ears, deep seated eyes, long philtrum and thin lips

**Discussion**

Sanjad-Sakati Syndrome is a very rare inherited autosomal recessive disorder that is distributed equally in both males and females. The gene location of this syndrome is on 1q42.3 chromosomes [3]. This syndrome is characterized by hypoparathyroidism, retardation of growth, and typical facial features, in addition to a variable degree of mental retardation. The most common abnormal physical findings of this syndrome are microcephaly, a narrow and long face, a beaked nose, deep-seated eyes, large ears, thin lips, a long philtrum, a small hand, and small feet [4],[5],[6],[7],[8]. Some of these features resemble Kenny-Caffey syndrome, DiGeorge syndrome, and familial isolated hypoparathyroidism, but the absence of a cardiovascular lesion, lymphocytopenia, or malformation of the skeleton makes it a different problem[9]. Ophthalmological assessments help us to differentiate between Sanjad-Sakati syndrome and Kenny Caffey syndrome. Corneal opacity and nanophthalmos are present in Kenny Caffey Syndrome patients, while these features do not describe in Sanjad-Sakati Syndrome patients apart from deep-seated eyes[10].

This syndrome is commonly described in people of Arabic origin in the Middle East; registered cases came from the kingdom of Saudi Arabia, Kuwait, Qatar, Oman, Morocco, and Jordan [11],[12],[13],[14],[15]. The first reported syndrome was by Sanjad et al. [2], then 12 cases were published in 1991 [4]: 6 boys and 6 girls. Symptoms also started in the neonatal period for 9 patients. All of these cases have hypoparathyroidism, severe hypocalcemia, and hyperphosphatemia. They also have dysmorphic features and growth retardation. There was no congenital heart disease in these cases, and cell-mediated immunity was normal in five of them. Kirk and Richardson reported 8 cases of them were brothers, the weight, height, and occipitofrontal circumference in these cases were less than -2SD for their ages. In all of these cases, typical facial features were presented. The first case involved relatives from Qatar in
the United Kingdom, while the others were from the Middle East region[11]. Marsden et al reported a 5-year-old girl from Saudi Arabia who presented with undetected parathyroid hormone, subnormal growth hormone. There is a normal renal response to parathyroid hormone, and growth hormone treatment results in a marked increase in weight and height. Hypocalcemia was controlled with 1-alpha-cholecalciferol [13]. A genetic study revealed that a Moroccan case with Sanjad-Sakati Syndrome presented with the Bedouin c.155-166Del mutation [14], also 6 of Jordan cases have the same deletion which resembles the mutation in our case[15]. This variant has previously been described as disease causing for Hypoparathyroidism-retardation-dysmorphic syndrome by Pal et al and Kerkeni et al [15],[16].

There is no particular finding in this case different from previously reported cases. Due to medical difficulties in making the diagnosis so we should have a high index of suspicion in order not to miss such cases. Because consanguineous marriages are common in our society, genetic counseling is crucial to lowering the risk of genetic mutation.

**Conclusion**

Although Sanjad-Sakati Syndrome is an incurable disease, early diagnosis helps with appropriate palliative treatment of the patient, reducing other associated metabolic and electrolyte disturbances, and allows for a genetic study and counseling of the family, especially in our society due to the high rate of consanguinity.

**Reference**

