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Original article

Sanjad Sackati Syndrome in Five Libyan Children: A Case Series

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Hypocalcemia, Hypoparathyroidism, Convulsion, Growth Retardation, Dysmorphism.

A B S T R A C T
Sanjad Sakati Syndrome (SSS) is one of the rare congenital disorders. It is characterized by congenital hypoparathyroidism, severe prenatal and postnatal growth retardation, dysmorphic features, and mild to severe intellectual retardation. Reported patients were almost exclusively from the Arabian Peninsula. We report on the first four cases (4girls) and one boy of Sanjad-Sakati Syndrome from the eastern part of Libya. They presented with recurrent attacks of hypocalcemic convulsions, severe growth retardation, and typical dysmorphic features of SSS. All patients came from consanguineous families. They all had a low PTH level, chest x-rays for cardiac size, and an echo were normal, so the diagnosis of Di George syndrome, Kenny-Caffey syndrome, is unlikely. This is the first report of Sanjad Sakati syndrome from Libya. To confirm the disease, there are plans to do a genetic study to provide proper counselling to the families. Only in the fifth case, after being diagnosed clinically, the genetic study was available, and the result was positive, as Sanjad-Sackati syndrome. If the disease is recognized early will lead to proper treatment of patients and prevent associated comorbidities. Pediatricians should consider this condition in the differential diagnosis of any dysmorphic hypocalcemia in children, especially newborns and young infants.

Introduction

HRD is an autosomal recessive disorder first reported in 1988 and confirmed by a definitive report in 1991 [1]. It has been reported almost exclusively in the Middle Eastern population and is characterized by congenital HY-hyperparathyroidism, retarded growth, mental retardation, seizures, and a characteristic dysmorphic feature. The molecular pathology of this syndrome was shown to be due to mutations in the TBCE gene in chromosomal area 1q42–q43 [2].

Features of Sanjad-Sakati syndrome include hypoparathyroidism with specific dysmorphic features, such as a beaked nose, deep-set eyes, a depressed nasal bridge, a long philtrum, microcephaly, micrognathia/retrognathia, thin lips, large floppy ears, and dental anomalies [3]. Consanguinity was present in most of the cases. Some had affected siblings who died in infancy. Increasing frequency of genetic disorders, particularly autosomal recessive conditions, is present in Arab populations owing to a high rate of consanguinity, large family size, and rapid population growth [4]. Di George syndrome and Kenny-Caffey syndrome have the same dysmorphic features of SSS. But SSS has no association with cardiac lesion, lymphopenia, or skeletal abnormalities, making it a distinct entity [5].

Case report

After taking their parents' consent, we are reporting five Libyan children with SSS (HRD) from the Benghazi children's hospital, Libya. A genetic test was not done due to a lack of facilities. To our knowledge, these are the first reported cases of SSS (HRD) from Libya

All five patients have growth retardation of variable degrees. Cardiovascular examination was normal in all cases. Laboratory investigation showed low total calcium, high phosphorus, normal level alkaline phosphatase, low or normal parathyroid hormone, which is essential for diagnosing hypoparathyroidism

Table 1 outlines five pediatric cases with striking similarities that suggest a hereditary disorder. All patients were born with low birth weight, showed symptoms within the first few months of life, and were diagnosed early, except for one delayed case. Each child experienced failure to thrive and came from consanguineous families, reinforcing the likelihood of a genetic condition. Family history reveals sibling connections and prior deaths with similar symptoms, further supporting a pattern of inherited disease. Overall, the data points to a consistent clinical presentation across related individuals, warranting genetic investigation and early intervention.

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Table 1. History of the 4 patients with Sanjad-Sakati syndrome

Variables	Case 1	Case 2	Case 3	Case 4	Case 5
sex	F	F	F	F	M
Birth weight	low	low	low	low	low
Age at onset of symptoms	2 months	45 days	4 months	37 days	17 days
Age at diagnosis	2 months	2 months	10 years	5 months	2 months
Failure to grow	+	+	+	+	+
consangunity	+	+	+	+	+
Family history	First baby in the family	She is the sister of the patient (case 3)	positive	One sibling died with similar symptoms	-

Abbreviation: F-female, M- male

Table 2 highlights a consistent pattern of physical and craniofacial abnormalities across five pediatric cases, all showing signs of microcephaly, deep-set eyes, beaked nose tips, thin lips, micrognathia, and small hands and feet. Most also have large, rotated ears. Despite these shared dysmorphic features and growth restrictions, all cases show normal echocardiographic findings, suggesting the condition primarily affects skeletal and facial development without involving the heart. The uniformity of traits points toward a likely genetic syndrome with a distinctive phenotype.

Table 2. Clinical findings of four patients with Sanjad-Skati syndrome

Variables	Case 1	Case 2	Case 3	Case 4	Case 5
Weight	3kg	3kg	10kg		3KG
Length	50 cm	48cm	69cm		46CM
Microcephaly	+	+	+	+	+
Deep-set eyes	+	+	+	+	+
Beaked nose tip	+	+	+	+	+
Big rotated ears	+	+	+	+	_
Small hands and feet	+	+	+	+	+
Thin lips	+	+	+	+	+
Micrognathia	+	+	+	+	+
Echocardiography	Normal	Normal	Normal	Normal	Normal

The lab results across all five cases reveal a consistent biochemical pattern: significantly low calcium levels and elevated phosphate levels, both of which fall outside normal ranges. Despite normal alkaline phosphatase activity, all patients show suppressed parathyroid hormone levels. This combination strongly suggests hypoparathyroidism, likely congenital and possibly genetic in origin, especially given the shared clinical features and consanguineous backgrounds previously noted. The findings support a diagnosis of a syndromic metabolic disorder affecting calcium-phosphate regulation.

Table 3. Results of biochemical and hormonal analysis

	Case1	Case 2	Case 3	Case 4	Case 5
Calcium N (9-11 mg/dl)	4.9mg/dl	5mg/dl	5.2mg/dl	4mg/dl	5mg/dl
Phosphate N (2.4-4.5 mg/dl)	7.2 mg/dl	8mg/dl	7.2mg/dl	7mg/dl	6.9mg/dl
Alkaline phosphatase N (55-260 IU)	164 IU	150 IU	200 IU	180 IU	220IU
Parathyroid hormone N (10-60 pg/ml)	4 pg/dl	5pg/dl	3pg/dl	5pg/dl	6pg/dl

Discussion

Hypocalcaemia should be considered in the differential diagnosis of any child presenting with afebrile convulsions. Hypocalcaemic convulsions may be clonic, focal clonic, multifocal, jacksonian, or apneic [4]. SSS is a rare autosomal recessive congenital disease. disease gene is on chromosome 1q42-q43 and has severe and often fatal consequences. It is commonly described in the Middle East population of Arabs [1].

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Sanjad-Sackati syndrome consists of hypoparathyroidism, which leads to hypocalcemic convulsion, intrauterine growth retardation, which continues throughout the whole life. Patient has typical dysmorphic features such as long, narrow face, deep set eyes, beaked nose, large floppy ears, long filiform, thin upper lip, and micrognathia. Patients have typical facial features and are mentally subnormal. Usually, they are presented early in the neonatal period as hypocalcemia, and investigations reveal a picture of hypoparathyroidism [6], reported patients were from Saudi Arabia, Qatar, Israeli Arab, Kuwait, Oman, Morocco, and Tunisia [8,2,7] and now we reported on five Libyan patients.
Our patients had a high rate of parental consanguinity. All of them have dysmorphic features with the same presentation of hyocalcemic convulsion in early months of life. All five cases have normal chest x-ray, normal echo, with these characteristic features and low parathyroid hormone, so diagnosis Sanjad Sakati syndrome was unrestrained, DiGeorge syndrome, and Kenny-caffey syndrome was considered unlikely.
A genetic study was not done because lack of facility(unfortunately), except for case 5, at which time a genetic study was available and the result was Ahomozygous pathogenic variant was detected in TBCE. We seem to agree with the explanation given by Tunisian authors as to the presence of Sanjad Sakti syndrome in the part of Africa was probably due to the arrival of Banu Hilal migrants with the spread of Islam to Africa in the 7th century 11.

Conclusion

We have reported the first case series of Sanjad Sakati syndrome in Libya. To confirm the disease, there are plans to arrange for DNA analysis to provide proper counselling to the families, and if the disease is recognized early will lead to proper treatment of patients and prevent associated comorbidities. Pediatricians should consider this condition in the differential diagnosis of any dysmorphic hypocalcaemic children, especially newborns and young infants. Early recognition of the disease will lead to proper treatment of patients and prevent associated comorbidities.

Conflict of interest. Nil

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