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Otolaryngologic Manifestations of Sanjad Sakati Syndrome- A Case Report

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ABSTRACT

Sanjad-Sakati syndrome (SSS) or hypoparathyroidism-retardation-dysmorphism (HRD) or Middle East syndrome is a rare autosomal recessive genetic manifestation seen predominently from the Middle East and Arabian Peninsula. Children affected with this condition are typically born with features of intrauterine growth retardation and present early with hypocalcaemic convulsions, typical facial dysmorphic features, severe growth retardation, developmental delay, low IQ and congenital hypoparathyroidism[1,2]. The condition is caused by mutations or deletions in the TBCE gene on Chromosome No.1 the locus is 230 kb region of gene with mutations in individuals who are affected[3].There are exceptional cases who are not affected due to a TCBE gene abnormality[4].

Keywords: Hypoparathyroidism, Facial dysmorphism, Recurrent ear infections

Introduction:

To date, fewer than 20 reports have been published worldwide in English discussing SSS. Overall, consanguinity was found in 2 of our 3 patients.

Patients with SSS typically present in the newborn period with tetany, seizures, or apnea due to hypocalcemia and recurrent infections, probably due to immune defects[5]. In the present study, most of the cases had been diagnosed during the neonatal period due to hypocalcemia/seizures or apnea, phenotypic pictures, or raised awareness of the syndrome among affected families. The syndrome has a wide variety of clinical features, including deep-set eyes, micrognathia, thin lips, small maxilla, severely decayed teeth, beaked noses, depressed nasal bridges, external ear anomalies, small hands and feet, short stature, and learning difficulties[6]. In addition, hypoparathyroidism and hypocalcemia are constant findings. All of our patients showed the cardinal features described previously

Neurological manifestations in the form of microcephaly, developmental delay, mental retardation, and seizures were reported in all patients. Speech was not affected grossly but they have some problems of articulation and they do well after speech therapy and they develop intelligible speech thereafter, some showed abnormal EEG tracings

Case report:

Here we present three children with following details who presented to us in ENT department of Sur Hospital, Oman multiple times as Outpatient with recurrent ear infections and upper respiratory infections. The following chart details the various features as below. This has been prepared based on computer retrospective records of case progress of these patients (Tables 1,2).

This syndrome has to be differentiated from Kenny-Caffey Syndrome which often shows cortical thickening of long bones with medullary stenosis, macrocephaly or normocephaly, normal mentality and immune deficiency. Kenny-Caffey Syndrome type 1 may show the same mutation in the tubulin specific chaperone-E gene[7] (Figure 1).

Patients' hypocalcaemia is usually treated with

Features	Case 1	Case 2	Case 3
Age	6 years	7 years	2 years
Date of Birth	2009	2008	2013
Sex	Female	Female	Male
Birth weight	1325 gms	1450 gms	1950 gms
Onset of symptoms	20 days after birth	18 days after birth	22 days after birth
Mental and motor development	Delayed milestones	Delayed milestones	Delayed milestones
Consanguinity	First degree	First degree	No
Micrognathia	Present	Present	No
Retrognathia	Present	No	No
Prominent forehead	Present	Present	No
Deep set eyes	Present	Present	Mild
Thin lips	Present	Present	Present
Depressed nasal bridge	Present	Present	Present
Beaked nasal tip	Yes	Yes	Yes
Low set ears(fig 1)	Present	Present	Normal ears
Big rotated ears	Yes	No	No
Microtia	No	No	Minimal
Lips	Thin lips with long philtrum	Thin lips	Thin lips
Tooth decay(fig 2)	Dental decay	Dental decay	Minimal decay
Palatal arch	Narrow	Normal	Normal
Ear infections	Recurrent pseudomonas otitis	Recurrent pseudomonas otitis	Recurrent AOM
Otoacoustic emission	Bilateral passed	Bilateral passed	Bilateral passed
Speech	Articulaton defect	Mild articulation problem	Delayed speech
Stertor	Mild	No	No
Latest weight	5500 GMS	6400 GMS	7200 GMS
Radiological findings	Nephrocalcinosis	Nephrocalcinosis	Normal
Calcium level N-2.3 to2.8 mmol/l	2.28	2.3	1.66
Phosphate level(n-1.45 to 2.16 mmol/l)	2.8	1.50	2.71
Pth (n-10-60 micromol/l)	<0.3	<0.1	<0.1
Creatinine	19.99 uMol/l	31.8 uMol/l	21.3uMol/l
Immunoglobulin levels	Normal	Normal	Normal
Chromosomal analysis	C.155-166 del 12 bp Homozygous	46 XX Normal Karyotype	46 XY Normal Karyotype
Others associations	Nephrocalcinosis, Hypothyroidism, Epilepsy, pinoscrotal hypoplasia and short penis	Labial fusion, vaginal atresia, Epilepsy	Epilepsy
Outcome of patient	Expired	Alive	Alive

Table 1: Computer retrospective records of case progress of these patients.

calcium and alpha calcidiol as we treated our patients. However they tend to continue having poor growth. Attempts to use growth hormone in some of the cases proved unsuccessful[8]. Most patients tend to die early with recurrent infections, but rare cases have survived up to the age of 18 years[9] (Figure 2).

Country	Number of reported cases	Clinical features	Genetic defect
Kuwait	21	Short stature, microcephaly, deep-set eyes, blue sclera, large floppy ear lobule, small hands and feet, mental retardation, hypoparathyroidism, and hypocalcemia	c.155-166del
Qatar	8	Dysmorphology, deep-set eyes, depressed nasal bridge with beaked nose, long philtrum, thin upper lip, micrognathia, large floppy earlobes, abnormal skeletal survey, developmental delay, hypoparathyroidism, hypocalcemia, and hyperphosphatemia	Not available
Belgium	2	Facial dysmorphism, short limbs, small hands and feet, small genitalia, hypoparathyroidism, and severe pre- and postnatal growth retardation	C.H.: c.66-67del c.1113T >A
Jordan	8	Short stature, microcephaly, deep-set eyes, small hands and feet, mental retardation and learning difficulties, hypoparathyroidism, hypocalcemia, hyperphosphatemia, and low levels of PT	c.155-166del
This report	3	Dysmorphism, preominent forehead, triangular face with deep-set eyes, abnormal external ear, wide depressed nasal bridge, bulbous nasal tip, anteverted nares, long philtrum, thin lips and downturned corners of mouth. The hands and feet were short, hypocalcemia, hyperphosphatemia, low levels of phosphorus	c.155-166del

Table 2: Comparitive review of genetic and clinical data of Sanjad Sakati Syndrome

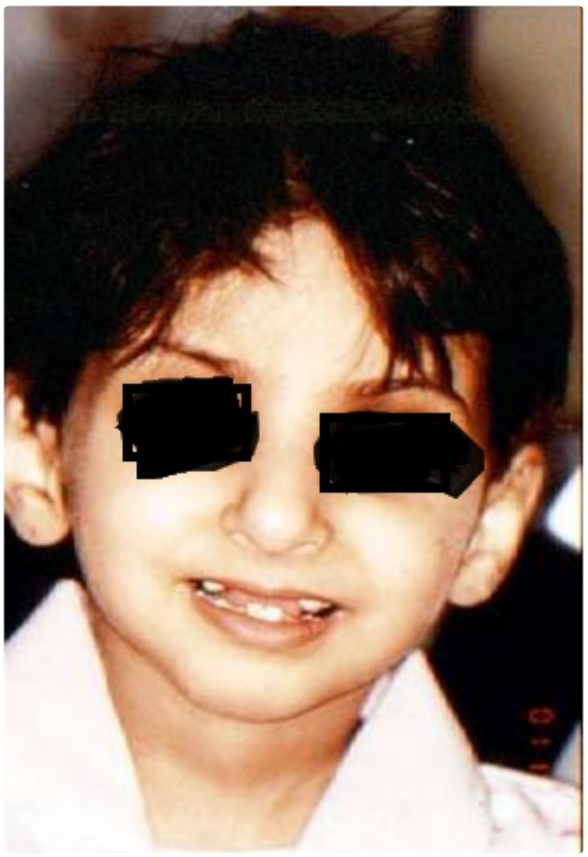

Figure 1: Kenny-Caffey syndrome type 1

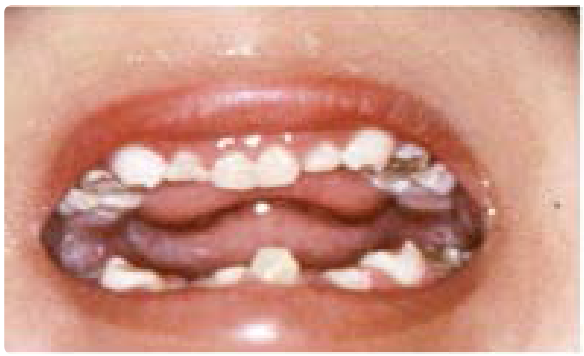

Figure 2: Patient Poor growth after treated with calcium and Alpha calcidiol

Conclusion:

SSS is a rare autosomal recessive disorder that is not uncommon in the Gulf and Arabian Peninsula. Most of the ENT manifestations are grossly present and children report due to various manifestation of poor immunity with chronically discharging ear, frequent rhinorrhea and upper respiratory infections. This case report details various facial and other clinical presentations involving SSS. Prevention of this syndrome may be achieved in the future through pre-implantation genetic diagnosis and carrier detection.

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