

## Sanjad - Sakati Syndrome in a Neonate

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Received: December 24, 2008;  
Initial review: February 26, 2009;  
Accepted: March 27, 2009.

Congenital hypoparathyroidism, growth retardation and dysmorphism is a rare autosomal recessive syndrome among Arab population commonly known as Sanjad-Sakati syndrome (SSS). Several metabolic and septic complications are known to manifest in the neonatal age. We describe the first report of morbid pathological fractures affecting a neonate with SSS.

**Key words:** Fracture, Neonate, Sanjad - Sakati syndrome.

Sanjad Sakati Syndrome (SSS) is a rare but well known entity described mainly in the Arab peninsula. The typical metabolic derangements lead to several morbid manifestations commencing as early as neonatal period and include hypocalcemia, seizures, nephrocalcinosis, increased susceptibility to infections, stunted growth and mental retardation. This report highlights an unusual occurrence of multiple longbone fractures in a neonate with SSS.

### CASE REPORT

A 34 week preterm boy (birthweight 1490 g) was born to a 19 years old Saudi lady (G<sub>2</sub>P<sub>1</sub>) by spontaneous vaginal delivery. Immediate neonatal period was uneventful. Baby passed meconium and urine on first day and was started on feeds. He developed fever and abnormal limb movements on 4<sup>th</sup> day associated with vomiting and clonic seizures in all four limbs. The child had facial dysmorphism in the form of microcephaly, deep set eyes, beaked nose, abnormal ear and micrognathia, short hands, and feet (**Fig. 1**). Investigations revealed hypocalcemia (Ca=0.9mmol/L), hypomagnesemia (Mg=0.45mmol/L); hypoparathyroidism (PTH=0.110 pmol/L; normal=1.59-6.89) and hyper-phosphatemia (2.25mol/L; normal=0.81-1.58). Septic screen

revealed normal CSF, but blood culture at 17th day grew *Klebsiella* sp. Baby was maintaining normal blood sugar and TORCH screening was negative. He was started on parenteral calcium, magnesium, pheno-barbitone and antibiotics. Feeding was regained on 20th day and baby started to tolerate supplemented milk. On 22nd day, he was noticed to have multiple abscesses over shin with fractures of both tibia and left humerus (**Fig. 2**). Retrospectively, it was found that fracture sites correlated with the sites attempted for IV placement by the NICU staff and a decision to place jugular Hickman catheter was taken to prevent further pathological fractures. 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 and pathological fractures took a long time (3 months) to heal.

### DISCUSSION

Sanjad Sakati Syndrome (SSS) or hypoparathyroidism-retardation-dysmorphism (HRD) syndrome is a rare but well documented autosomal recessive syndrome predominantly seen in Arab peninsula (1). It is characterized by congenital hypoparathyroidism (hypoPTH), prenatal and



FIG. 1 Features of Sanjad Sakati Syndrome (deep set eyes, abnormal ear, beaked nose, microcephaly, short feet).

postnatal growth retardation, seizures and a 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. Metabolically, babies suffer from often severe and fatal hypocalcemia, hypomagnesemia, hyperphosphatemia and congenital permanent hypoPTH. These metabolic derangements are responsible for nephrocalcinosis, medullary stenosis of long bones 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, reports of a variant without TBCE mutation has also been documented(2).

The phenomenon of multiple pathological fractures have not been reported in SSS scenario so far. Although medullary stenosis due to thickening of cortex in SSS has been documented, osteopenia and pathological fracture in early neonatal age in SSS is

intriguing. In adults, PTH is considered anabolic to trabecular bone and catabolic to cortical bone. Hypoparathyroidism leads to positive growth of cortical bones and variable effects on trabecular bones usually measured by bone densitometry(3). However, bone densitometry studies are deficient in assessing mineral content and stress bearance aspect of bones carrying both the trabecular and cortical types and this study was not conducted in our child.

We conclude that pathological fractures could complicate SSS in the neonatal period. Adequate mineral supplemented milk, radiological survey of the skeleton and delicate handling of the limbs particularly during lifting and placing IV access are some of the precautions that may prevent such morbidity. Bone densitometry is recommended to identify babies at risk of such complications.

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FIG.2 X-ray long bones showing pathological fractures.