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Case Report

SANJAD–SAKATI SYNDROME- RARE SYNDROME ASSOCIATED WITH RECURRENT PSEUDO-OBSTRUCTION – A CASE REPORT

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ABSTRACT
Sanjad-Sakati syndrome (SSS) or Middle East syndrome is one of the rare syndrome with varied features and presentation. We are presenting a case report of this syndrome with particular features of recurrent pseudo-obstruction-making the pediatric surgeon and radiologist alert all the time for further management. Metabolic disturbance which is causing pseudo-obstruction in this child has to be kept in mind to prevent unnecessary surgical management.

Keywords*: Sanjad–Sakati Syndrome, Hypocalcaemia, Pseudo-Obstruction*

INTRODUCTION
Sanjad-Sakati syndrome (SSS) is also known as hypoparathyroidism-retardation-dysmorphism (HRD) or Middle East syndrome. It is one of the rare genetic disorders that have been reported predominantly in the gulf region including the Saudi Arabia, Oman and Kuwait. It is autosomal recessive (Ahmed Farag *et al.,* 2013) disorder and is running in families with consanguineous marriage. Arab tribes who had more history of consanguineous marriage had more incidence of Sanjad-Sakati syndrome than rest of the world. This syndrome had spectrum of varied clinical, biochemical, radiological, dermatological and morphological disorders.

The initial report of Sanjad *et al.,* in 1988 and its definitive report in 1991, clearly established this as a distinct disorder comprising of congenital hypoparathyroidism, hypocalcaemia, hyper-phosphataemia, severe intrauterine and postnatal growth failure, respiratory infection susceptibility, dwarfism, mild to moderate mental retardation, seizures and, abnormal dentition (Hussein *et al.,* 2014).

Morphological feature of this syndrome (Abdul Cader *et al.,* 2016) includes 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. Dermatological features includes calcinosis cutis (Hussein *et al.,* 2014).

CASE
It is a case of a pre-term boy child born via vaginal delivery with the weight of 1350grm, out of consanguineous marriage. On birth the child showed respiratory distress and developed recurrent respiratory tract infection. Neurologically, the child had cerebral irritability and features of birth asphyxia, which shows significant neural- development as the child matures. Predominant GI symptoms included feeding intolerance, abdominal distension and sever gastroesophageal reflux. Metabolically child had hypocalcemia, hypo-magnesimia, and hypo para-thyroidisim and metabolic alkalosis. Hypocalcemia is resistant to treatment. In addition to the multisystemic features of SSS, our case had a unique presentation of recurrent pseudo- obstruction which is managed conservatively.

DISCUSSION
Synonyms
Sanjad-Sakati syndrome (SSS) / hypoparathyroidism-retardation-dysmorphism (HRD) / Middle East syndrome/ Hypoparathyroidism-intellectual disability-dysmorphism syndrome/ Hypoparathyroidism-short stature-intellectual disability-seizures syndrome/ Richardson-Kirk syndrome.

Case Report

Incidence and Prevalence

Unknown (Rare Disease-OMIM 241410)). It is among the rare disorders, in which, there are isolated case reports found in the literature, most reported in the gulf regions and Arab speaking area in Africa.

Mode of inheritance

It is autosomal recessive inheritance with gene locus corresponding to lq42-43 (Kelly *et al.,* 2000). 42 cases had reported up to March 2016 **(**Segana Hasan *et al.,* 2016**).** In most of the cases, parents are healthy heterozygous carries, child acquire by means of inheritance (25%).

Radiological features

This case presents with the feature of left renal atrophy or agenesis, with compensatory hypertrophy of the right kidney (Figure 1). Right kidney shows normal Doppler study of the renal vessels. These features had not been reported in any other literatures. But this child also has a feature of bilateral undescended testes (Figure 2), which has been reported in one among the three cases reported by Mona Hafez *et al.,* in Egypt in March 2017. There are no features of medullary sclerosis (Figure 3) seen in this case, as compared to the case reports series published from the Egypt by El hassanien *et al.,* in 2013. This may be varied presentation or the medullary sclerosis can develop in the later age of the child.

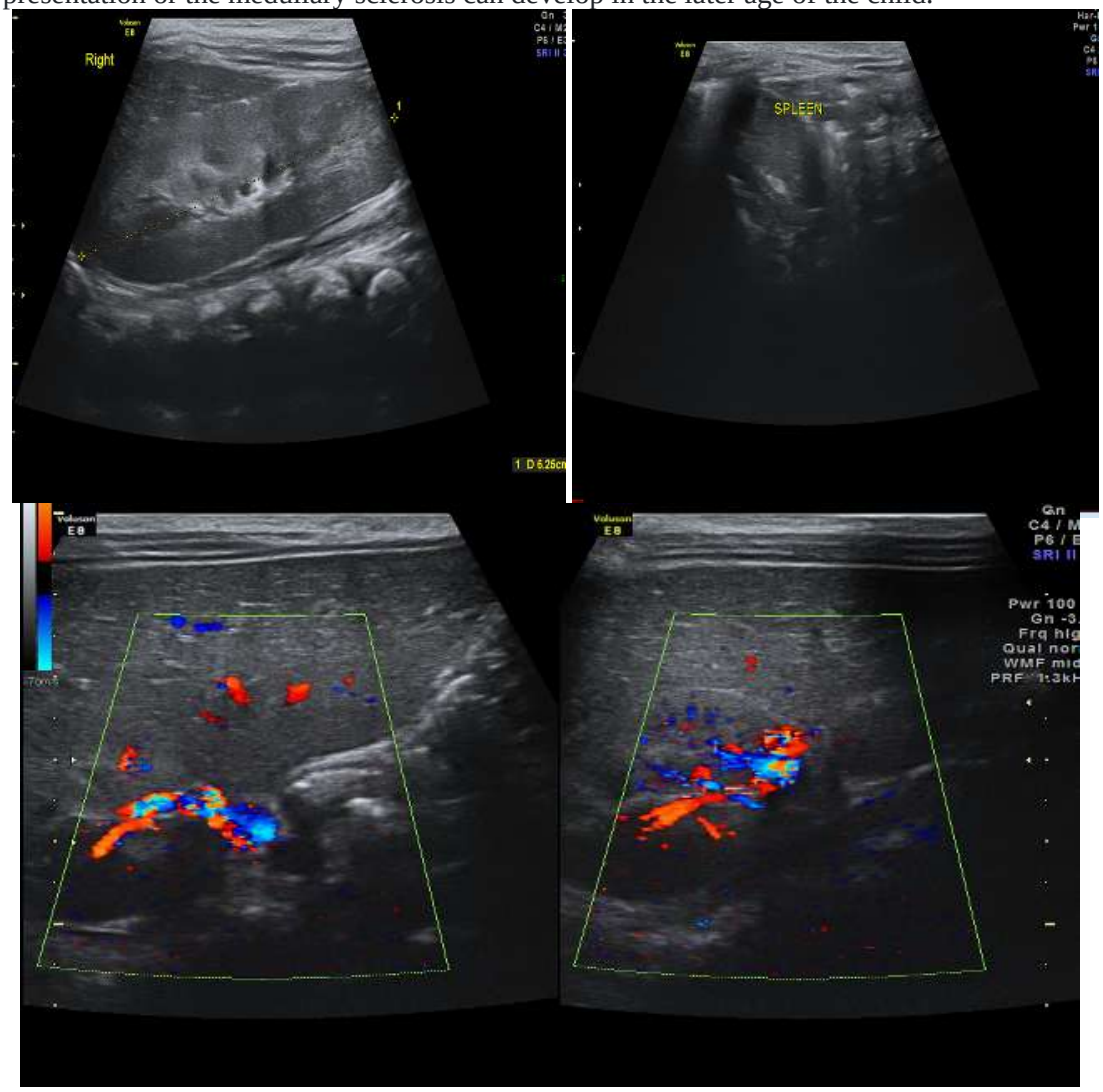
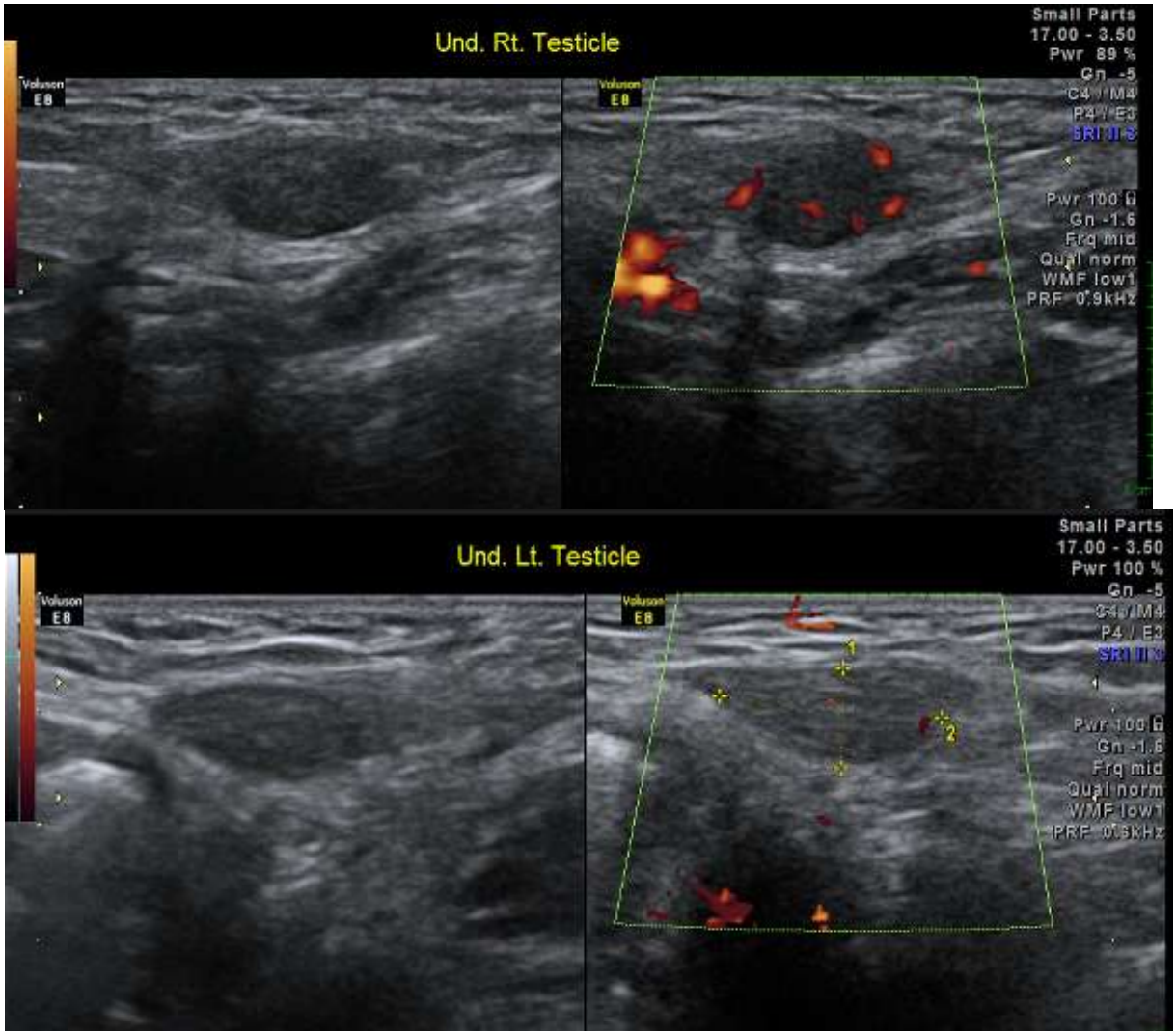


Figure 1



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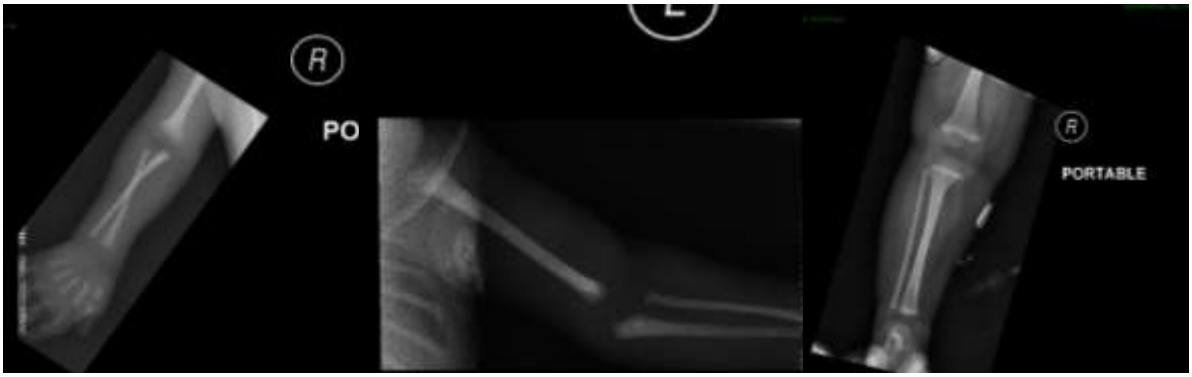

Figure 3

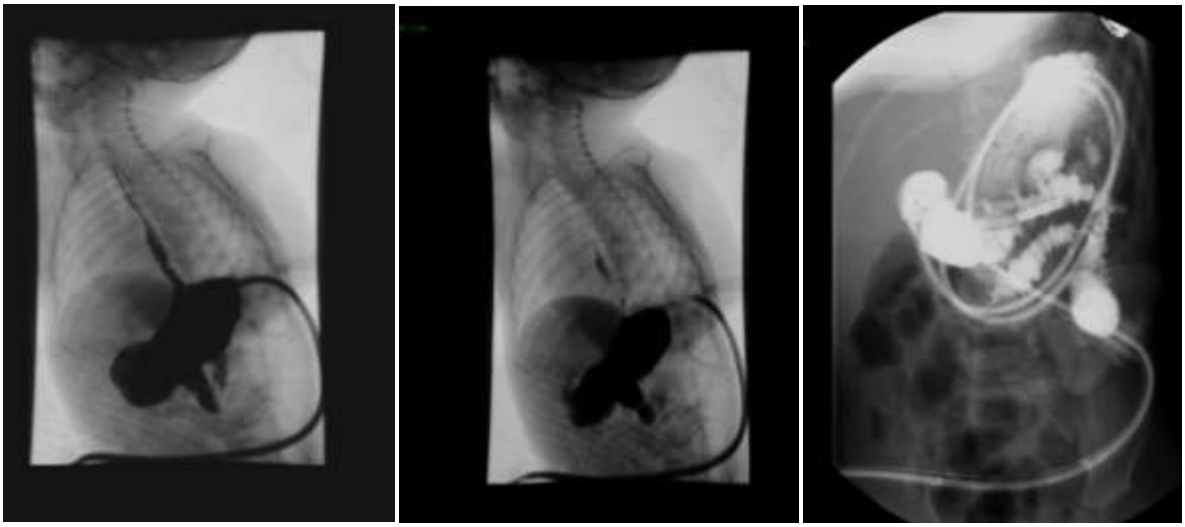

Figure 4

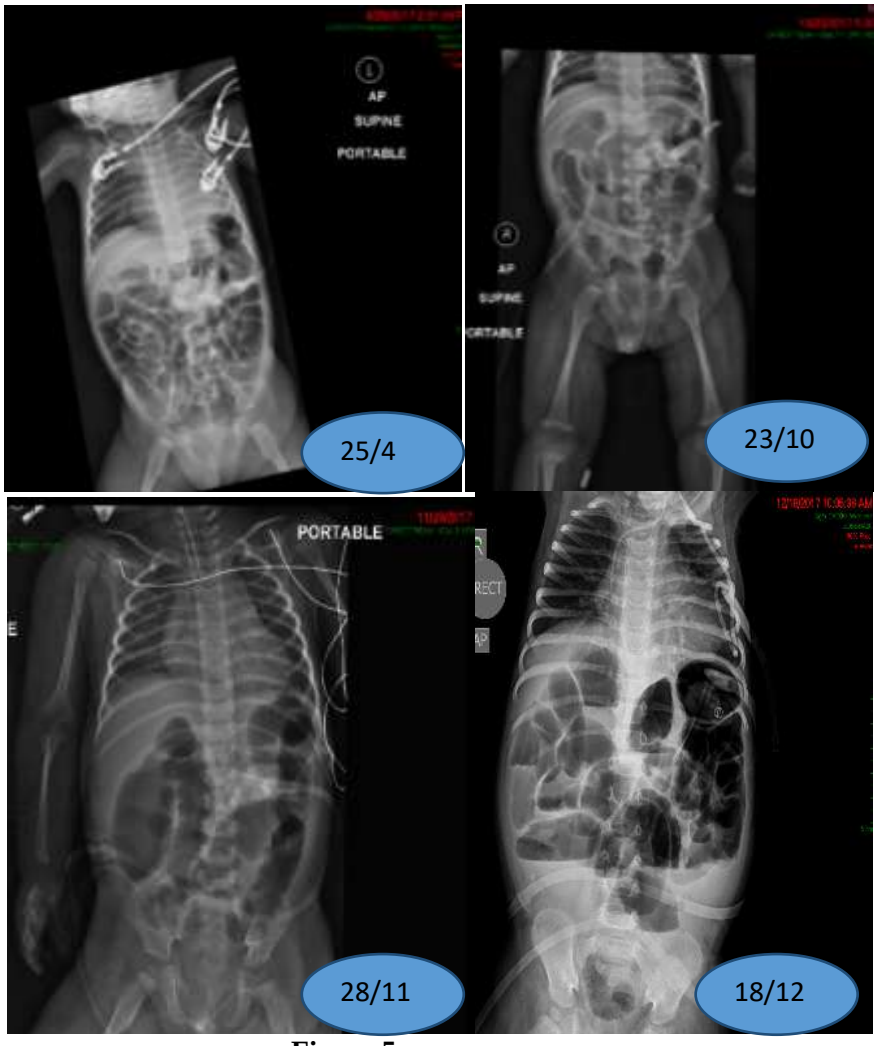

Figure 5

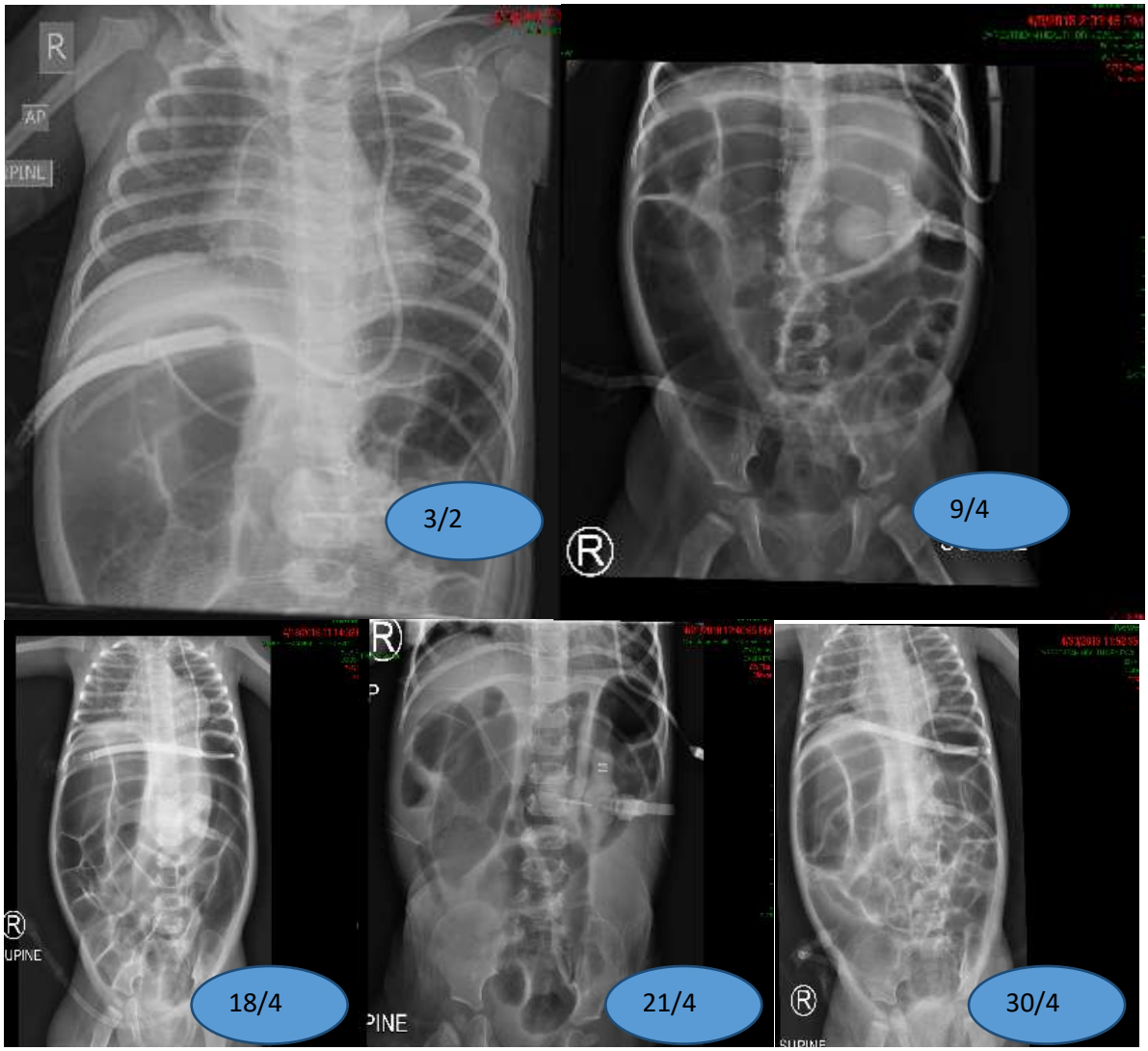

Figure 6 (2018)

Gastrointestinal features

This child had presented with sever feeding intolerance with significant GE reflux and vomiting. To improve the feeding, gastrostomy tube was placed. Fluoroscopy image through the gastrostomy tube shows significant GERD (Figure 4) with reflux of contrast up to the cervical esophagus. Presence of GERD has not been found in previous literatures.

Another unique feature of this case report is recurrent pseudo-obstruction. This child is showing recurrent pseudo-obstruction since birth (Figure 5 and Figure 6). There were repeated admissions due to recurrent abdominal distension, which may or may not be associated with metabolic disturbance. This persisted or was treated after admission with or without the correction of metabolic causes. There is no definite association found between the metabolic cause and recurrent large bowel dilatation, one of the possible causes is visceral myopathy causing chronic intestinal pseudo obstruction as described by Pal *et al.*, in 2010.

Conclusion

Sanjad–Sakati syndrome –The syndrome associated with multi systemic multi spectrum of features, association of Severe GERD and recurrent pseudo-obstruction had to be kept in mind for proper management of multi systemic syndrome.

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