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Arora-Nirwane-Samant syndrome: A variant of Freeman-Sheldon syndrome

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Abstract
Freeman-Sheldon syndrome (FSS) is a rare inherited form of distal arthrogryposis characterized by craniofacial abnormalities, camptodactyly with ulnar deviation of the fingers and talipes equinovarus. We present a case variant of FSS with proximal and distal arthrygryposis with the additional features of bilateral developmental dysplasia of the hip and bilateral congenital vertical talus deformity.

Keywords: Arora-Nirwane-Samant Syndrome; Freeman-Sheldon Syndrome; distal arthrogryposis; development dysplasia of hip; congenital vertical talus

1. Introduction
Freeman-Sheldon syndrome (FSS) is a rare inherited form of distal arthrogryposis characterized by craniofacial abnormalities, camptodactyly with ulnar deviation of the fingers and talipes equinovarus. Less than 100 cases of the disease have been reported in the literature. We present a case variant of FSS with proximal and distal arthrygryposis with the additional features of bilateral developmental dysplasia of the hip and bilateral congenital vertical talus deformity. We also review the literature on FSS.

2. Case Report
BK was a 2 yr old male brought to our institution with multiple soft tissue contractures and bilateral rocker-bottom feet limiting his activities of daily living. These features were present since birth and were non-progressive in nature. The prenatal, perinatal and post-natal history was insignificant for underlying risk factors.
On examination, the patient had frontal bossing, open fontanelles, microstomia, poor dental hygiene, micrognathia and short neck (**Figure 1**). The patient had no hearing difficulties.

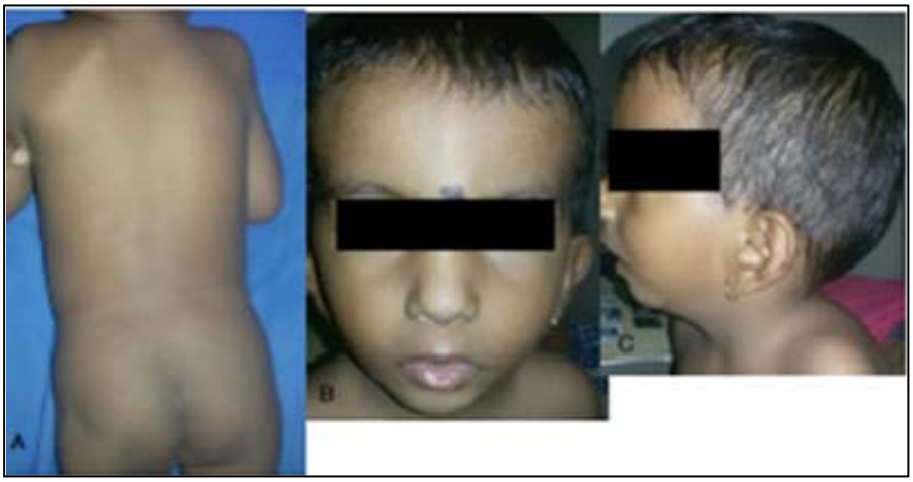

Fig 1: (A) Clinical photograph of patients back and pelvis; (B) clinical photograph of the front of patient face; and (C) clinical photograph of the side of patient's face

The upper limbs (**Figure 2**) revealed an adduction and internal rotation deformity of both shoulders, the right elbow was in a fixed flexion contracture of 90 degrees and the left elbow in a fixed flexion contracture of 30 degrees. This was acceptable with the Indian activities of daily living whereby the right hand is used for feeding and the left hand for personal hygiene. Bilaterally the hands exhibited flexion contractures of the long flexors of the fingers and bilateral thumb in palm deformities suggestive of adduction contractures.

Fig 2: (A) Clinical photograph of the right upper limb; (B) clinical photograph of the left upper limb; (C) lateral radiograph of the right elbow; (D) lateral radiograph of the left elbow; (E) clinical photograph of right hand; and (F) clinical photograph of left hand

The trunk (**Figure 1**) exhibited mild kyphoscoliosis with a barrel chest and decreased respiratory excursion. The bilateral hips exhibited fixed adduction and internal rotation deformities. Bilaterally (**Figure 3**), the knees had a fixed flexion deformity of 10 degrees and the foot was in bilateral congenital vertical talus with deep dorsolateral and posterior skin creases.

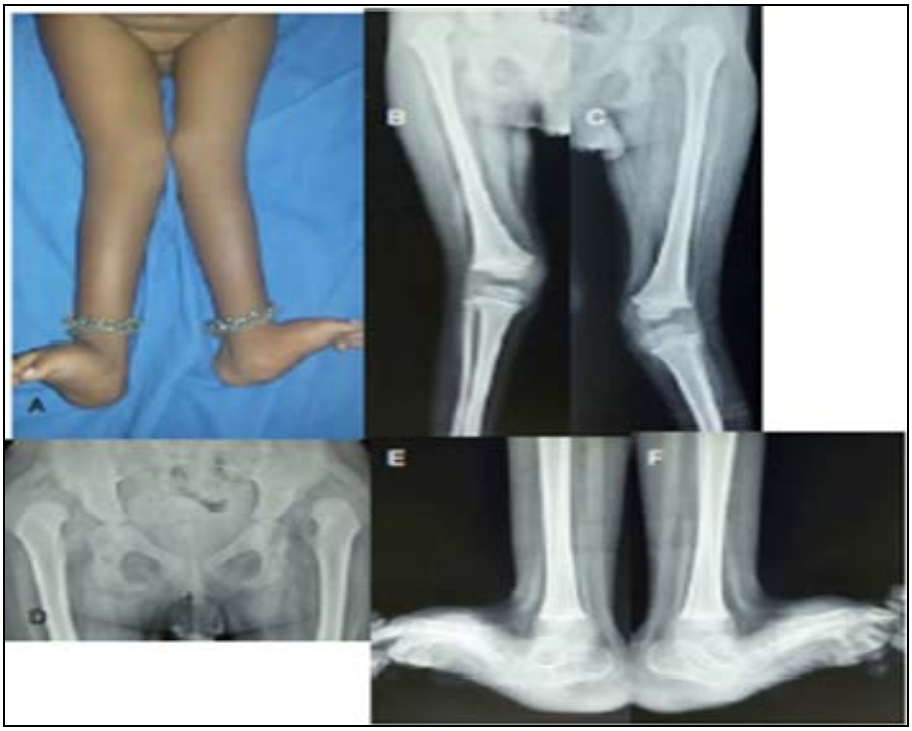

Fig 3: (A) clinical photograph of the bilateral lower limbs; (B) Anteroposterior radiograph of the right knee; (C) Anteroposterior radiograph of the left knee; (D) anteroposterior radiograph of bilateral hips; (E) lateral radiograph of right foot; and (F) lateral radiograph of left foot

Radiographs were suggestive of mild kyphoscoliosis of the spine, bilateral DDH with formation of false acetabula (**Figure 3**), bilateral genu valgum and bilateral CVT. Scrotal and abdominal ultrasound revealed undescended testis in the abdominal space.

3. Discussion

Freeman-Sheldon syndrome (FSS) is a rare inherited form of distal arthrogryposis characterized by craniofacial abnormalities, camptodactyly with ulnar deviation of the fingers and talipes equinovarus. It was first described in 1938 by Freeman and Sheldon [1]. It is also known as Windmill-Vane-Hand syndrome, cranio-carpo-tarsal dysplasia and whistling face syndrome. Although most cases (70%) occur sporadically it is thought to be transmitted by autosomal dominant inheritance with variable penetrance [2]. Less than 100 cases of the disease have been reported in the literature.

Cranio-facial abnormalities are multiple. The increased tone and fibrosis of the facial muscles gives rise to an immobile mask-like facial expression. In addition to hypertelorism, the eyes are deeply set below a supraorbital ridge with blepharophimosis and ptosis. The nose is small and the alae nasi are hypoplastic. The myopathic fibrotic circumoral musculature results in microstomia with characteristic protruding pursed “whistling” lips and a long prominent philtrum. Contracted facial musculature also results in micrognathia [3].

Common orthopaedic manifestations include kyphoscoliosis, pectus excavatum, spina bifida and intercostal myopathy. In the upper limb, ulnar deviation of the wrist and flexion contracture of the fingers occurs. In the lower limb, talipes equinovarus, vertical talus and contracted toes may be present.

The main dental feature of FSS is severe microstomia which limits access for routine dental care [4]. Patients suffer from anesthetic problems, with difficult intubation and intravenous access cited in the anesthetic literature [5].

The precise mechanism of combined skeletal and muscular dysplasia is unclear, although Sauk et al. suggested that the hypoplasia of the muscle bundles supplied by the motor branch of the major nerves may cause these abnormalities [6]. Biopsy of the affected muscles reveals fibrosis which may contribute to the contractures. Recent studies focus of mutations in the MYH3 gene as a cause for this syndrome, suggesting that the congenital contractures are a shared outcome of prenatal defects in myofiber force production [7]. Biopsies of the affected facial muscles show atrophy of the muscular fibers with abundant infiltration of adipose tissue, fibrosis, central migration of the nucleus and variations in the diameter of muscular fibers [8].

In contrast to the aforementioned features, our syndrome varies in that the arthrogryposis is suggestive of amyoplasia (proximal and distal arthrogryposis), as opposed to distal arthrogryposis. Further, our child exhibited congenital vertical talus as opposed to the more common congenital talipes equinovarus. Further in contrast to the ulnar deviation of the fingers, our child displayed thumb in palm deformities bilaterally. There was also the additional attribute of bilateral developmental dysplasia of the hip with development of false acetabula, and bilateral undescended testis.

4. References

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