Freeman-Sheldon Syndrome – A Case of Rare Observation

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Abstract
A two months ten days female child of non consanguineous parents without family history of the disease presented with watering from both eyes since birth, cough and respiratory distress for three days. She has dysmorphic mask like facies which has whistling appearance and other features include deep set eyes, broad nasal bridge, short neck, hypoplasia of the ala nasai, long philtrum, small tongue, high arched palate with “H” shaped dimple on chin. There is contracture deformity of knee, toes and finger. The diagnosis of this rare disease FSS is made incidentally accordingly, counseling and treatment of associated illness is given. We should be aware of this disease although rare and proper counseling is given to the parents to prevent future complication from this disease.

Case
Sabina, two months ten days old female child of non consanguineous parents admitted in our hospital with the complaints of watering from both eyes since birth, cough and respiratory distress for three days. She had no significant past history of any illness. She was born by normal delivery at 39 weeks gestation and it was uneventful. Mother had no history of miscarriages and illness during pregnancy. She is the third issue of her parents and other siblings are normal. On examination, the child face is dysmorphic mask like facies with a small mouth and pursed lip having the appearance of whistling, deep set eyes with epiphora, broad nasal bridge, short nose, hypoplasia of the ala nasai, long filtrum, short tongue, high arched palate, “H” shaped dimple on the chin and mid face. She also has short neck (Figure 1). There is contracture of both knees. Flexion contractures of both hands and toes of the right foot are found (Figure 2). Anaemia, jaundice, cyanosis, clubbing and oedema are absent. Temperature 98, heart rate 140/minute, Respiratory rate 56/minute, weight three kilogram, weight for age Z score -3.2SD, length 52 cm, weight for length Z score -2.8SD, occipito frontal circumference 35.5 cm (below third percentile).

On respiratory system examination, respiratory rate is 56/minute. Chest in drawing is present. Crepitation is found in all zones of both lung fields. Other system reveals no abnormality.

X-ray chest is suggestive of nonspecific pulmonary infection. X-ray of the upper and lower limb revealed incurving of fingers

Figure 1,2: Showing dysmorphic features in Freeman-Sheldon syndrome
of hand and flexion position of knee joints (Figure 3). The child is treated with anti-infective agents for respiratory illness. Finally genetic counseling and future advice about the disease is given.

Discussion

Our case is diagnosed on the basis of medical history and physical examination that reveals characteristic dysmorphic status, combining bone anomalies and joint contractures with typical facies. Female sex is affected from this disease as shown by the majority of case reports [1,3]. Our observation is similar to others. We find microcephaly in our patient which is not mentioned in other reports. There is no history of consanguinity in our case like other observations which suggests sporadic cases due to mutation only in the proband. Respiratory complications are common and life threatening in these patients due to structural anomalies of the oropharynx and upper airways [5]. Our case also has respiratory difficulties which are due to respiratory infection that might be associated with structural respiratory anomalies.

Most reported cases of FSS occur sporadically with no family history of the disease, though there are reports of a specific pattern of either autosomal dominant or recessive inheritance. A variant of FSS, DA2B (MIM601680) is caused by mutations in TNN12 encoding troponin 1 [6]. The prototypic DA, DA1 (MIM 108120), is caused by mutations TPM2 [7,8]. The mechanism(s) by which mutation in TPM2 and TNN12 cause multiple congenital contractures is unclear [5]. This syndrome should be differentiated from other arthrogryposis syndrome, Schwartz-Jampel syndrome and Trismus-pseudo camptodactyly syndrome [3]. The prognosis or natural history in these children is feeding difficulty, vomiting and dysphagia leading to failure to thrive. Most of the features are secondary to increased muscle tone. Aspiration is the important cause of early mortality in these cases. Majority of who survive with moderate motor and speech delays in childhood usually have normal intelligence and life expectancy. Dental crowding and oral hygiene secondarily to small mouth can develop. There post natal growth deficiency, anesthetic complications are relatively common.

![Figure 3: X-ray of the upper and lower limb revealed incurving of fingers of hand and flexion position of knee joints.](image)
i.e., before surgery they are to be carefully evaluated [3]. Genetic counseling has to begin to the parents explaining the nature, the prognosis and the management of this syndrome. Recurrence risk in the future offspring is probably slightly higher than the population risk [3]. It is impossible to carry out prenatal diagnosis through direct DNA analysis. Ultrasonographic evaluation can help the prenatal diagnosis of FSS as it was reported for 20 weeks fetus with a positive family history [9]. It revealed abnormalities of the extremities and mouth.

There is no standard management protocol. The foot and hand abnormalities are resistant to treatment and require conservative and operative measures. Multiple, extensive orthopedic and plastic reconstructive surgery is often required. Surgical correction of microstomia is important from both aesthetic and functional points of view (food intake especially). We have advised our patient to have surgical consultation whenever there is difficulty in feeding due to microstomia. Parents are also suggested to feed semisolid and non sticky foods which are easily swallowed and to keep the mouth of the child clean thus protect from dental caries. Structural anomalies of the oropharynx and upper airways are a constant concern for general anesthesia. Tracheal intubation via direct laryngoscopy often cannot be carried out. The spine should be preoperatively to perform epidural/spinal anesthesia. Intravenous access may be difficult because of limb deformities and thickened subcutaneous tissues. Patients may be at increased for malignant hyperthermia. The follow-up of these pediatric patients requires prolonged orthodontic and orthopedic treatment including speech and hearing [5].

References