



### Freeman-Sheldon Syndrome: About a Rare Observation

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#### ABSTRACT

Freeman-Sheldon Syndrome (FSS), also known as Distal Arthrogryposis Type 2A (DA2A), Craniocarpotarsal Dystrophy, Windmill-Vane-Hand Syndrome, or Whistler Face Syndrome, was first described by Freeman and Sheldon in 1938 [1,2]. It is a very rare syndrome of multiple congenital contractures characterized by a microstomy giving the appearance of the "whistling face", a club foot and articular contractures. FSS is the most severe form of distal arthrogryposis. We report a rare observation of a two years old child, who has a poly malformative syndrome admitted in our ENT department at the Specialty Hospital, Rabat. On examination, the child presents a facial dysmorphism with head bird facies with prominent forehead, hyperthelorum and small mouth. Otherwise, H aspect of the chin and mandibular retrognathia are noted. At the endoral examination, we notice a cleft palate, dental overlap and glossoptosis. Moreover, osteo-articular examination shows hypertonicity of the limbs, especially on the fingers, and a left club foot. The geneticist's opinion is in favor of a Freeman-Sheldon syndrome. The child has benefited from a surgical closure of the cleft palate. FSS is caused by genetic abnormalities involving the "distal arthrogryposis multiplex congenita" gene, whose inheritance may be either mostly autosomal dominant or autosomal recessive. The management by health professionals of several medical disciplines must be undertaken from birth to evaluate the severity of their phenotypic presentation and minimize the risk of patient development delay.

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#### Introduction

Freeman-Sheldon Syndrome (FSS), also known as Distal Arthrogryposis Type 2A (DA2A), Craniocarpotarsal Dystrophy, Windmill-Vane-Hand Syndrome, or Whistler Face Syndrome, was first described by Freeman and Sheldon in 1938 [1,2]. It is a very rare syndrome of multiple congenital contractures characterized by a microstomy giving the appearance of the "whistling face", a characteristic face, a club foot and articular contractures. FSS is the most severe form of distal arthrogryposis.

#### Material and Methods

We report the clinical observation of a two years old child, who has a poly malformative syndrome admitted in our ENT department at the Specialty Hospital, Rabat. The child is born of a not followed pregnancy without notion of medication taken by the mother. There is also a notion of first degree consanguinity. The history of the disease dates back to birth by a difficulty in breastfeeding with suction, swallowing and ventilation disorders. In addition, the mother reports a notion of difficult feedings, fake roads, ebb feeding through the nasal cavities and a contracture of the four members. On examination, the child presents a facial dysmorphism with a bird head facies and a prominent forehead. There is also hyperthelorum with a small mouth, H-aspect of the chin and mandibular retrognathia (Figure 1, 2). Otherwise, at the endobuccal examination, we notice a cleft palate, a class 2 dental relation, an overlap of teeth and a glossoptosis. On osteo-articular examination, there is hypertonicity of the limbs, particularly on the fingers, and a left club foot (figure 3).



Figure 1. Hyperthelorum with small mouth, H-aspect of the chin and mandibular retrognathia.



Figure 2. Bird's head Facies with prominent forehead.

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**Figure 3. Contracture of the limbs especially to the fingers.**

A paraclinical assessment was performed thus the CT of the facial mass showed a retrognathism, a narrowing of the upper parts of choana with cleft palate and a slight filling of the posterior ethmoidal cells. Otherwise, the rest of the paraclinical examination including ENT, ophthalmic and cardiac is normal.

The geneticist's opinion is in favor of a Freeman-Sheldon syndrome.

The child has benefited from a surgical closure of the cleft palate.

#### Discussion

Freeman-Sheldon syndrome is a type of distal arthrogryposis associated with distal type 1 arthrogryposis (DA1). In 1996, more precise diagnostic criteria made it possible to classify the disease as distal arthrogryposis type 2A (DA2A) [1,2]. "DA1" is the least severe arthrogryposis. Then comes the DA2B being more severe than DA1 by presenting additional symptoms that respond less to therapy. The most severe being DA2A (Freeman-Sheldon Syndrome) that is most resistant to therapy because of its multiple symptoms [3-5]. As a result, Freeman-Sheldon Syndrome has been described as a type of congenital myopathy.

In March 2006, Stevenson et al. have published a strict diagnosis to identify the syndrome. This diagnosis includes two or three symptoms of distal arthrogryposis: microstomy, "whistling face", dimples in H on the chin. Symptoms of this syndrome include raised eyelids, strabismus, low ears, a long Philtrum, gradual hearing loss, scoliosis and walking difficulty. Gastroesophageal reflux has been observed in childhood, but usually it resolves as the child gets older. The tongue may be small, and the limited movements of the soft palate may cause rhinolalia. Often, there is an appearance of dimples in the shape of "H" or "Y" on the chin [4].

FSS is caused by genetic abnormalities so Krakowiak et al. have decoded the "distal arthrogryposis multiplex congenita" gene, a very similar syndrome, in phenotypic expression, from classical FSS, to 11p15.5-pter [6]. Other mutations have been found [7]. Heredity can be either autosomal dominant (most often demonstrated) [8-10], or autosomal recessive [11-14].

Patients should seek maxillofacial and orthopedic surgery advice as soon as possible in order to improve their functions or for aesthetic purposes [15-17]. Operational measures must be carefully pursued, avoiding radical measures and taking into account the abnormal physiology of FSS-related muscles. Unfortunately, many surgical operations produce results inferior to optimal results, due to myopathy syndrome. When surgery is necessary, it should be done as early as possible taking into account the fragile health of FSS

patient. Early intervention minimizes the risk of delayed mental development and avoids long-term rehabilitation [18].

#### Conclusion

The FSS is a rare type of distal arthrogryposis, but generally clinically recognizable. This rare syndrome inheritance may be either autosomal dominant or autosomal recessive. Patients with FSS should be evaluated and managed from birth by health professionals from multiple medical disciplines to assess the severity of their phenotypic presentation and to minimize the risk of patient developmental delay.

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