Freeman-Sheldon Syndrome

A 12-week-old male child presented with deformities of the feet and hands. On examination the patient had a “whistling mouth” facies, microstomia, broad nasal cartilage and deep set eyes (Fig. 1). There was ulnar deviation of the hands, cortical thumb and flexion of fingers (Fig. 2). The child had bilateral clubfeet. There were no other visceral malformations. On the basis of the typical clinical features a diagnosis of Freeman Sheldon Syndrome was made.

The syndrome is primarily an autosomal dominant condition, though a few autosomal recessive types have been reported. It is a type of distal arthrogryposis with most of the features being secondary to increased muscle tone. These children are recognized by the characteristic facies and the associated joint deformities. Microcephaly and mental retardation have been reported in about a third of these subjects. Dysphagia in early infancy may contribute to failure to thrive and death due to aspiration. Treatment includes managing deformities of the hands and feet, speech and dental anomalies. Long term outcomes have been reported to be satisfactory in the large majority of these patients.

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