The Turkish Journal of Pediatrics 36: 329-332 1994

WHISTLING FACE (FREEMAN-SHELDON) SYNDROME IN TWO SIBLINGS*

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SUMMARY: Bekir N, Bayraktaroğlu Z, Coşkun Y, Karaaslan C. (Departments of Ophthalmology and Pediatrics, Gaziantep University Faculty of Medicine, Gaziantep, Turkey). Whistling face (Freeman-Sheldon) syndrome in two siblings. Turk J Pediatr 1994; 329-332.

Two siblings with typical manifestations of whistling face (Freeman-Sheldon) syndrome (WFS) born to unaffected parents are presented. In Case 1, deep-set eyes, epicanthus, blepharophimosis, right lid ptosis, strabismus, anti mongoloid slant, small mouth, mask-like face, high-arched palate, nasal speech, dysphagia, kyphosis and minimal scoliosis were noted, while Case 2 displayed blepharophimosis, mask-like face, long philtrum, high-arched palate, scoliosis, bilateral post-axial polydactyly of the feet and pes varus. We corrected the blepharophimosis in Case 1 by bilateral canthotomy and canthoplasty.

This syndrome is usually inherited as an autosomal dominant trait; however, some authors have reported an autosomal an autosomal recessive form of this syndrome similar to our cases. Nevertheless, this could be explained by genetic expression of the mutant gene. *Key words: whistling face syndrome, blepharophimosis, canthotomy, canthoplasty, genetic heterogeneity.*

This disorder was described by Freeman and Sheldon in 1938[1]. The most common manifestations of Freeman-Sheldon syndrome are midface flatness with small mouth giving a "whistling" appearance, deep-set eyes, epicanthal folds, strabismus, ulnar deviation of fingers, blepharophimosis and talipes equinovarus[2].

Although the occurrence of autosomal dominant inheritance has been clearly established, the recessive form of this syndrome has yet to be definitively evaluated[3].

We describe here two siblings with whistling face (Freeman-Sheldon) syndrome (WFS) born to unaffected parents, and we present our surgical approach to lid deformities.

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Case Reports

The phenotypically normal parents with consanguinity (marriage between cousins) had four children, two of them unaffected. The mother and father were 27 and 29 years old, respectively.

Case 1

This seven-year-old boy had typical manifestations of WFS (Fig. 1): deep-set eyes, epicanthus, blepharophimosis, right lid ptosis, strabismus, anti-mongoloid slant, small mouth, mask-like face, high-arched palate, nasal speech, dysphagia, kyphosis and minimal scoliosis. Vertical lid distance of the right eye was 6 mm for downward gaze and 7 mm for upward gaze, while horizontal lid distance was 22 mm. Vertical lid distance of the left eye was 8 mm for downward gaze and 9 mm for upward gaze; horizontal lid distance was 23 mm. Lateral canthotomy and canthoplasty were performed on the patient's lids.

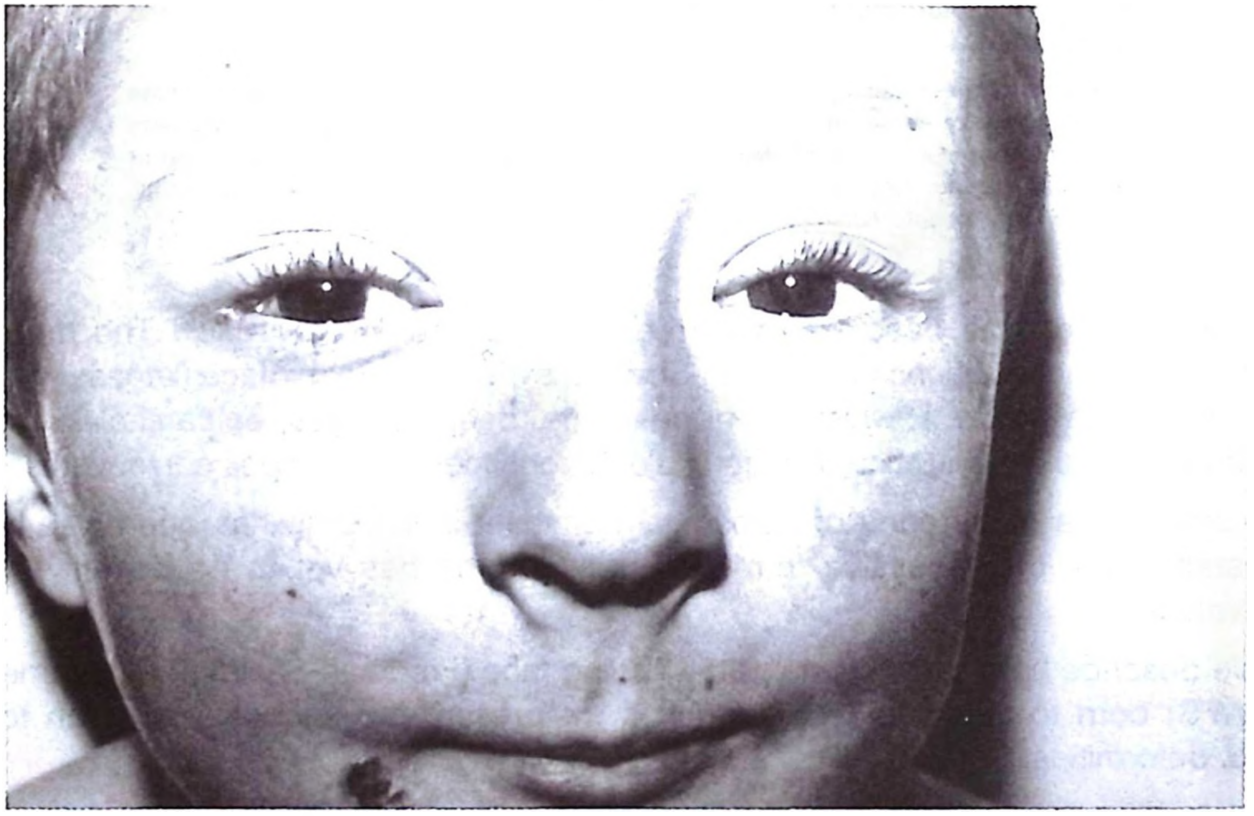

Fig. 1: The typical "whistling face" appearance of Case 1.

Case 2

The three-year-old sister of Case 1 (Fig. 2) had abnormal features of extremity and face which included blepharophimosis, mask-like face, long philtrum, high-arched palate, scoliosis, bilateral post-axial polydactyly of the feet and pes varus. Vertical lid distance of the right eye was 6 mm for downward gaze and 7 mm for upward gaze, with a horizontal lid distance of 20 mm. Vertical lid distance

of the left eye was 7 mm for downward gaze and 8 mm for upward gaze; horizontal lid distance was 21 mm. In this case polydactyly of both feet was correted.

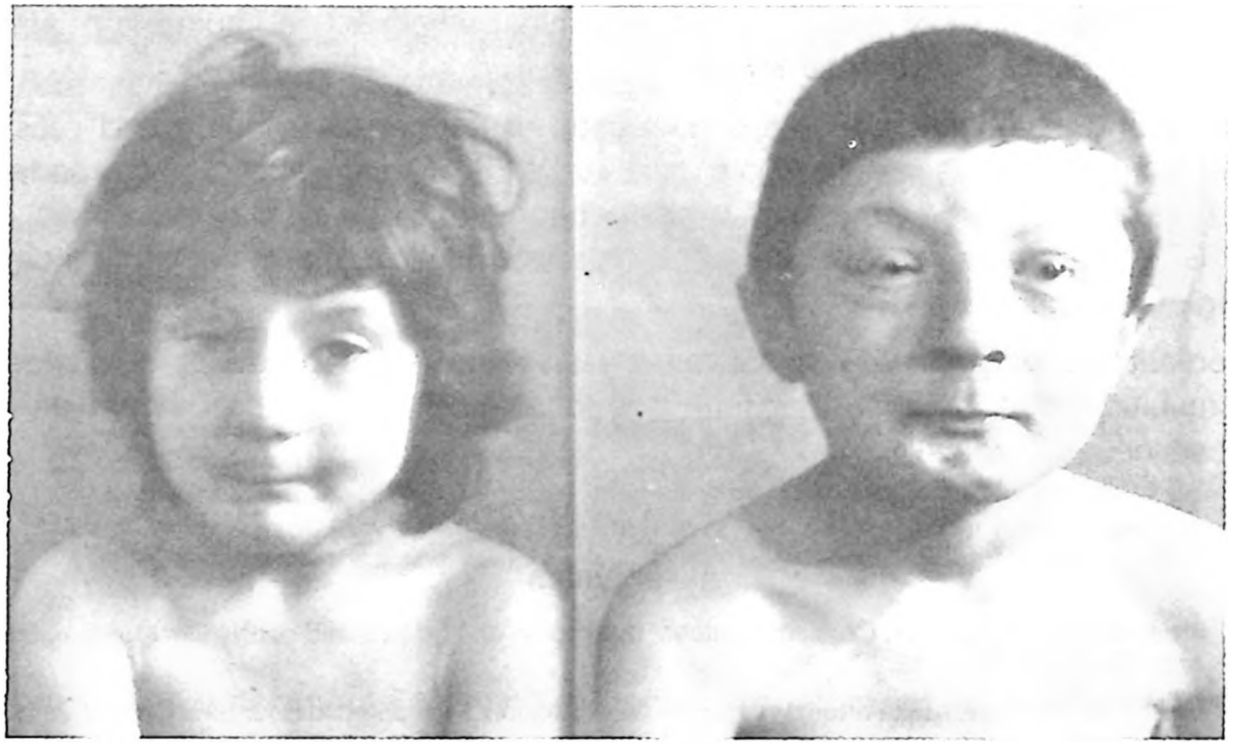

Fig. 2: Photograph of Case 2.

Discussion

Patients born to normal consanguineous parents often have characteristic manifestations of WFS[1,4]. Although there is general agreement in the literature that genetic inheritance of WFS is autosomal dominant[3], some reports suggest an autosomal recessive transmission of this syndrome[5-8]. The two siblings reported here are likely to have had autosomal recessive inheritance because of consanguineous unaffected parents; however, we found no clue in differentiation of their phenotype and clinical severity of the disease from patients with the autosomal dominant from. This evidence suggests that genetic heterogeneity including variable expressivity of the mutant gene or gonodal mosaicism can be seen in WFS, as in our cases. Some authors reported similar cases, indicating an autosomal recessive form of WFS, but they did not observe phenotypical and clinical differences from the autosomal dominant form of the disease[8,10]. In conclusion, since there is no agreement regarding the occurrence of autosomal recessive inheritance in WFS, it is more convenient to consider this form of genetic transmission in practical implications and genetic counselling. A new aspect of WFS was reported by Vanek et al.[11] They observed two cases of WFS with severe abnormalities of the extremities but only slight anomalies

of the face and claimed that the primary cause of the deformities was related to myopathy, classifying this syndrome as a separate type of myopathic arthrogryposis. Bilateral post-axial polydactyly of the feet in Case 2 is an unusual finding in this syndrome; however, it is unclear whether this finding is related to WFS or is a coincidental matter.

Guyuron and Winkler[12] reported correction of congenital upper eyelid ptosis and bilateral commissuroplasty in a case with WFS. Guzzanti et al.[13] also reported orthopedic aspects concerned with the locomotor apparatus in two cases of WFS. We corrected the blepharophimosis in Case 1 by bilateral canthotomy and canthoplasty.

Medical treatment for WFS is not available, and prenatal diagnosis of the disease is insufficient at present. It is beneficial to correct some of the deformities of these cases surgically.

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