FREEMAN – SHELDON SYNDROME: A CASE REPORT

O. Güven, M.D ** / M. Başaran, M.D. * / A. Pamukçu, M.D. ***

* *Associate Professor, Department of Pediatrics, Faculty of Medicine, Marmara University, İstanbul, Turkey.*
** *Assistant Professor, Department of Orthopedics and Traumatology Faculty of Medicine, Marmara University, İstanbul, Turkey.*
*** *Research Asistant, Department of Pediatrics, Faculty of Medicine, Marmara University, İstanbul, Turkey.*

SUMMARY
A case of Freeman - Sheldon syndrome with typical facial appearence and bone deformities is reported. Additional abnormalities such as mitral insufficiency, sensoryneural hearing loss, cerebral and cerebellar atrophy are also noted.

Key words: Freeman - Sheldon syndrome, cranio-carpo-tarsal dysplasia.

INTRODUCTION

In 1938 Freeman and Sheldon described a syndrome in two children with deep set eyes, ocular hypertelorism, whistling mouth appearence, marked ulnar deviation, flexion contractures of the fingers and bilateral talipes equinovarus deformity (1). High arched palate, flexion contractures of the toes, nasal speech, kyphoscoliosis, convergent strabismus, growth retardation with normal intelligence, and contractures of the hips with difficulty in abduction were also reported in the following years (2, 3, 4, 5). The familial occurence has been noted and an autosomal dominant inheritance pattern was suggested (6).

Because of extreme rarity of this syndrome we report this case and compare the findings with previous reports.

CASE REPORT

A 14 year old girl (H. K.) was admitted to the hospital because of deformed extremities, inability to talk and walking - difficulty.

History revealed that deformities was first recognized soon after birth and flexion contractures became more severe with time. Past medical history was otherwise unremarkable. The father and mother were first degree cousins. The elder sister suffered from mental and motor retardation of undetermined etiology. Two other siblings were healthy.

Physical examination: Her weight (21 kg), height (120 cm) and Head Circumference (49 cm) were all below 3 rd percentile. She was unable to hear and talk. Convergent strabismus, deep set eyes, Long Filtrum typical whistling mouth appearence were noted. Cardiac auscultation revealed grade III/VI pan-systolic ejection murmur at apex with axillary transmission. Breast development was at stage III according to Tanner's classification. There was no pubic or axillary hair.

In orthopedic examination bilateral internal rotation of shoulders, flexion contractures and muscular atrophy at hips (Figure 1), thoracal scoliosis between T_2 - T_7 with the angle of 35 degree towards left side (Figure 2) were found. There was radial deviation and irregular flexion contractures of fingers (Figure 3).

The feet showed bilateral "pes planovalgus" deformity. There was extreme extension contracture of the first toe and severe flexion contracture of the second toe of the right foot preventing her to wear shoes (Figure 4).

Neurologic examination was normal. The intelligence could not be evaluated due to hearing loss but she seemed to be appropriately responsive to social contacts.

Laboratory Findings: Audiography showed bilateral sensoryneural deafness. Blood biochemistry and hematologic tests were all within normal limits. Bone X - Rays confirmed thoracal scoliosis and finger contractures with osteoporosis (Figure 5). CT scan of the head showed cerebral and cerebellar atrophy (Figure 6) and echocardiography revealed congenital mitral insufficiency.

DISCUSSION

Freeman - Sheldon syndrome or Cranio-Carpo - Tarsal dysplasia is an extremely rare disorder. Thus, only few cases appeared in literature for the past 50 years. Owing to its rarity the full spectrum of findings, possible variants and inheritance pattern are not very well documented.

However whistling mouth appearence, deep set eyes, severe flexion contractures of the fingers particularly in thumbs, vertebra deformities, growth retardation, and talipes equinovarus were repeatedly reported (2, 3, 4, 5, 6).

In our case, characteristic facial appearence, scoliosis, flexion contractures of the fingers and toes, growth retardation, contractures of the hips with limited abduction were consistent with previous reports. On the other hand, we found radial deviation of the hands rather than ulnar deviation. Talipes equinovarus was absent but there was pes planovalgus deformity.

Additionally a congenital mitral insufficieny, sensoryneural hearing loss, cerebral and cerebellar atrophy and delayed puberty were present. These findings are not mentioned in previous cases suggesting a more severe form of the syndrome.

Although familial occurence has been noted and autosomal dominant inheritance pattern was suggested (6), parental consanguinity was absent in other cases.

In our case mother and father were second degree relatives. The elder sister was suffering from a mental and motor retardation according to the history. It was not unfortunately possible to examine her.

Chromosomal studies are planned in the family members in order to show possible abnormalities.

The surgical correction of deformities was of limited success. The contractures of the hands and feet cannot be treated by soft tissue operations because of high incidence of recurrences. Therefore a metatarsophalangeal arthodesis was performed to the right foot in order to allow the patient to wear shoes which improved walking.

REFERENCES

1. *Freeman EA, Sheldon SH. Cranio - Carpo - Tarsal Dystrophy. An undescribed congenital malformation. Arch Dis Child. 1938; 13: 271 - 283.*
2. *Otto FMG. Die "Cranio - Carpo - Tarsal Dystrophie" (Freeman Sheldon). Ein Kasiustisclar Beitraq 2. Kinderkeilk. 1953; 73: 240 - 250.*
3. *Külz J. Das Freeman - Sheldon - Syndrom. Med Bild. 1961; 4: 79 - 81.*
4. *Burian F. The Whistling Face characteristic in a compound Cranio - Facio - Corporal syndrome. Brit J Plast Surg. 1963; 16: 140 - 163.*
5. *Weinstein SS, Corlin RS. Cranio - Carpo - Tarsal Dysplasia or the Whistling Face syndrome. Amer J Dis Child. 1969; 117: 427 - 433.*
6. *Keselstein EG, Abrahamov A. Familial occurence of the Freeman - Sheldon Syndrome: Cranio - Carpo - Tarsal Dysplasia. Pediatrics. 1971; 47 - 6: 1064 - 1067.*

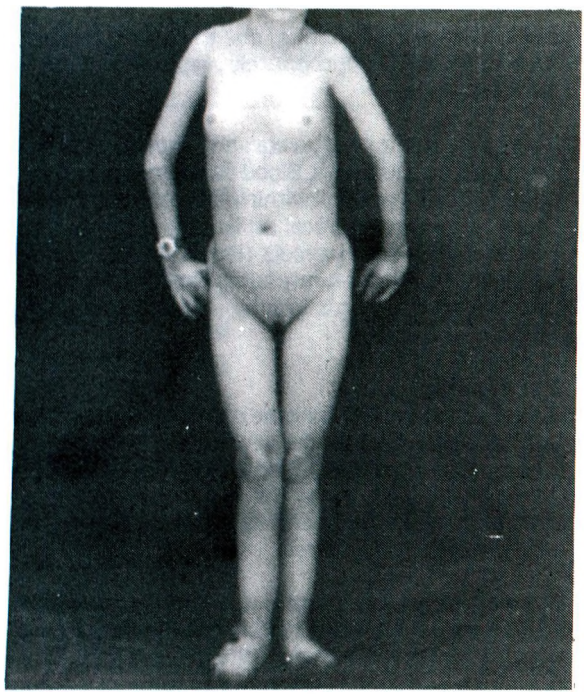

Fig. 1.

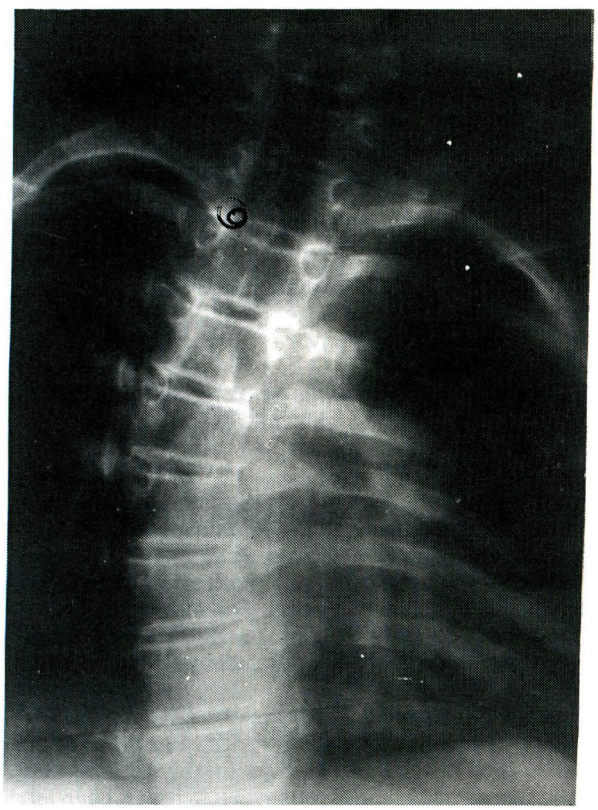

Fig. 2.

Fig. 3.

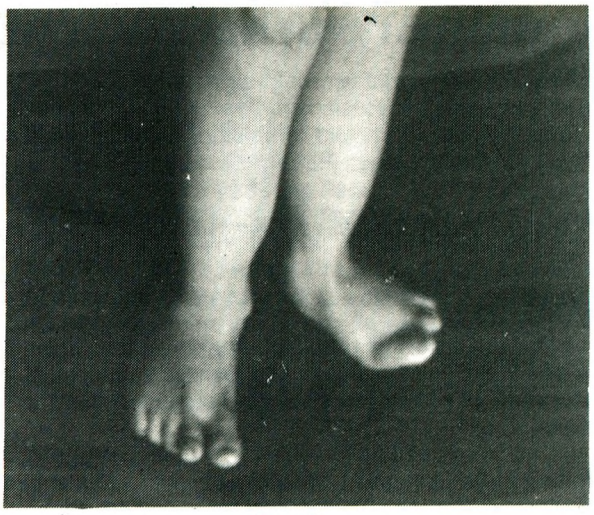

Fig. 4.

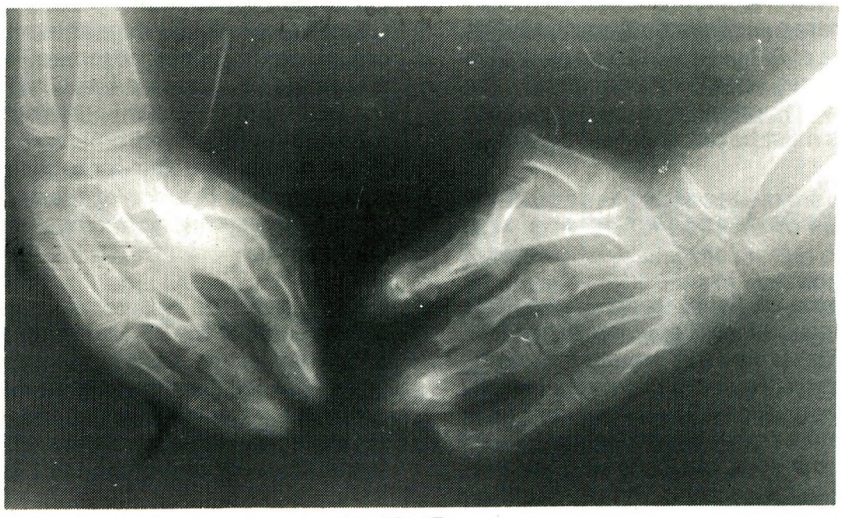

Fig. 5.

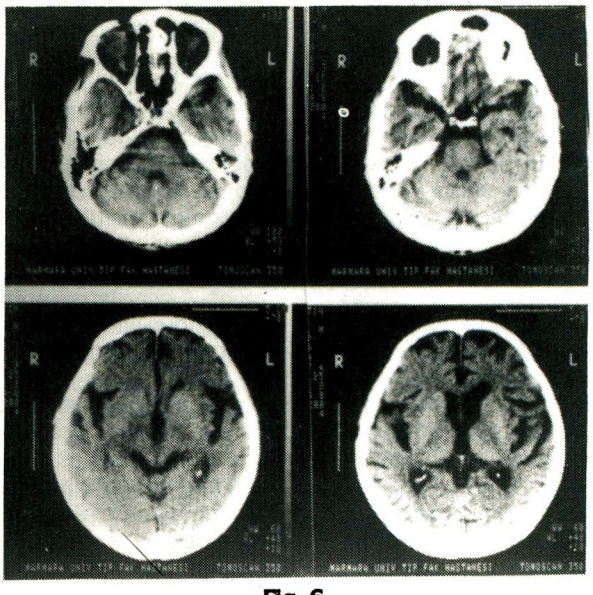

Fig. 6.