LARSEN'S SYNDROME WITH MIXED-TYPE HEARING LOSS

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Mixed-type hearing loss is an extremely rare feature of Larsen's syndrome, a complex of symptoms characterized by multiple joint dislocations and „flat" faces. In this report, a patient with bilateral mixed-type hearing loss and Larsen's syndrome is presented.

Keywords : hearing loss ; Larsen's syndrome.
Mots-clés : surdité ; syndrome de Larsen.

INTRODUCTION

Larsen's syndrome was initially described in detail by Larsen *et al.* in 1950 (4). However, the earliest case was reported by Mc Farland in 1929 (6). The syndrome is characterized by large joint dislocation, flat faces and hypoplastic nails (3).

Although in most of the cases autosomal recessive inheritance has been observed, an autosomal dominant trait is also reported (1, 3, 5, 6). In addition, Stanley *et al.* claim that most of the cases occur sporadically (7).

Since 1990, 50 cases of Larsen's syndrome have been reported (2, 5). Mixed-type hearing loss was present in only one of these cases (8).

CASE REPORT

C. Y. is a 13-year-old boy, whose parents are first cousins. The mother had not been exposed to known teratogens during her pregnancy. The family history reveals that there are 3 healthy siblings and no relatives with orthopedic problems or hearing loss.

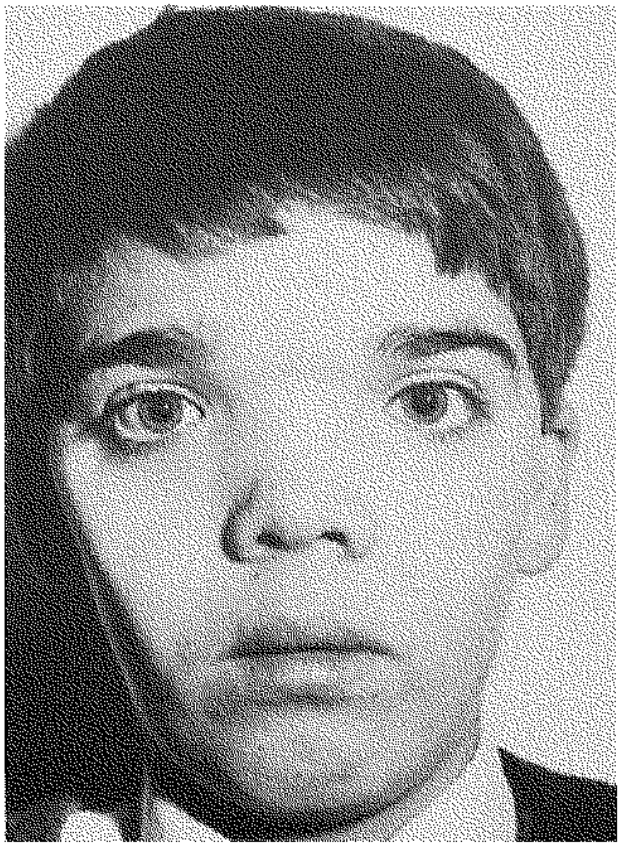

Fig. 1. — The face of the patient on physical examination, characterized by flat face, prominent forehead, hypertelorism, and depressed nasal bridge with anteverted nostril.

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The patient's height was 139 cm and head circumference was 52 cm. A flat face, prominent fore head, hypertelorism, depressed nasal bridge, anteverted nostril and high palate were present on physical examination (fig. 1).

Orthopedic examination revealed a volar subluxation of the wrists, dislocated elbows and hips, and bilateral talipes equinovarus. There were ulnar and palmar deviation of the hands and atrophy of the dorsal lumbrical muscles. All the fingers were long but not cylindrical in shape. Short nails, spatulate thumbs and a simian line were present on the left hand (fig. 2).

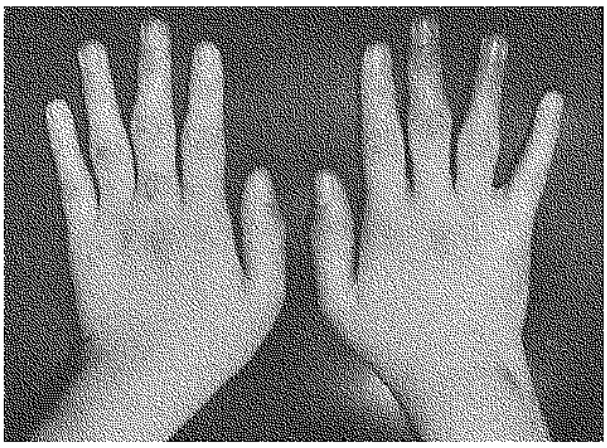

Fig. 2. — The view of the spatulate thumb and fingers which do not show the characteristic cylindrical shape.

Skeletal radiographs confirmed the clinically evident dislocations. Besides a flattened fifth cervical vertebral body, hypertrophic synostosis at the transverse processes of the sixth and seventh cervical vertebrae and left hemivertebrae were found between sixth and seventh cervical vertebrae (fig. 3). Mild thoracic scoliosis was convex to the right.

Hand radiographs showed an excessive number of carpal bones bilaterally, a large and deformed lunatum and multiple ossification centres at both proximal and distal ends of the first, second, third, and fifth metacarpal bones bilaterally (fig. 4). Radiographs of both feet demonstrated calcaneal deformation.

Otoscopic examination revealed normal tympanic membranes. However, the results of the pa-

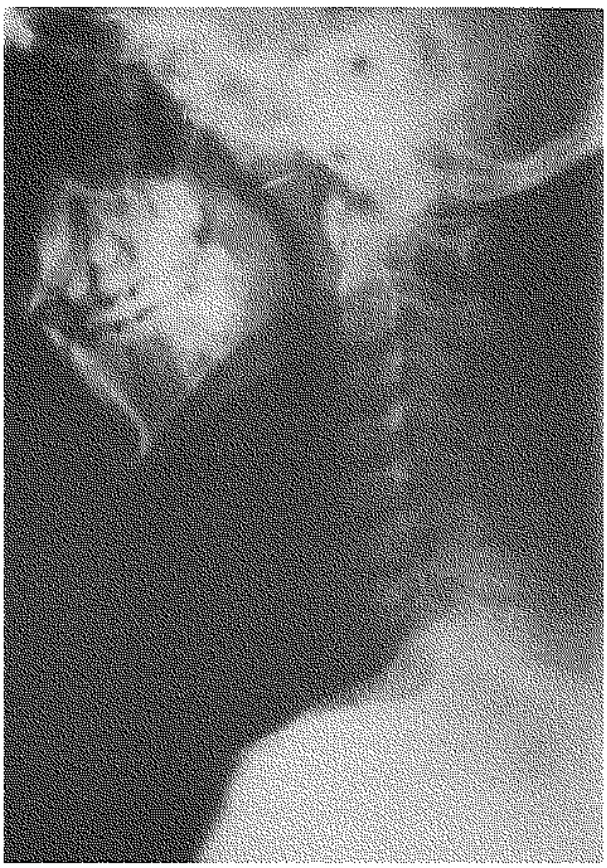

Fig. 3. Flattened fifth cervical vertebral body and hypertrophic synostosis at transverse process of the sixth and seventh cervical vertebrae.

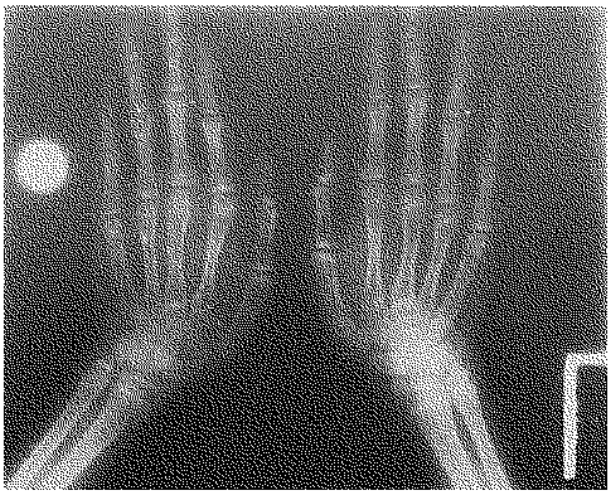

Fig. 4. — Excessive number of carpal bones and multiple ossification centers of the carpal bones.

tient's audiogram and tympanogram demonstrated mixed-type hearing loss in both ears. The audiogram revealed a hearing loss of 23 db for the air canal and 15 db for the ossicular canal in the left ear. These values were 30 db and 13 db respectively in the right ear. The tympanogram confirmed the results of the audiogram.

A few serial operations (iliac osteotomy, derotation and varisation at the age of 2 years, Achilloplasty, medial release, and posterior capsulotomy at the age of 4 years and bilateral cuboid enucleation at the age of 6 years, were performed in order to reconstruct the foot and hip deformities. During the reconstructive operation of talipes equinovarus it was observed that the deformity in the calcaneus prevented the correction of the equine position. The patient is still wearing an orthopedic boot.

DISCUSSION

Steel and Kohl describe extra ossification centres in the calcaneus (8). Delayed ossification, elbow dislocation, abnormal segmentation of vertebrae, and excess number of carpal bones are also described in this syndrome, originally described in 1950 by Larsen (4). Additional features such as mental retardation, congenital heart defects, hydrocephaly (4) and orofacial abnormalities (6) are also reported. In 1976, a case of Larsen's syndrome with otitis media in association with cleft palate defect was reported (7).

In 1988, Stanley *et al.* reported a child with Larsen's syndrome who had mixed-type hearing loss. They discussed the pathogenesis of hearing loss observed in their patient and in three patients previously reported by Hermann *et al.* (1981), Renault *et al.* (1982), and Ventruto *et al.* (1976) and concluded that either recurrent otitis media or joint dislocations of middle ear ossicles seen in Larsen's syndrome are the underlying factors (8).

The presence of ossicular abnormalities is supported by the occurrence of an incudostapedial joint abnormality observed by Maack and Muntz in one of their patients with Larsen's syndrome. Mixed-type hearing loss was found on routine examination in our patient.

In our 13-year-old patient hearing loss possibly resulted from the malformation of ossicular bones, as there were no ear complaints or history of otitis media.

One of the characteristic features of Larsen's syndrome, nontapering cylindrical finger, was not present.

This syndrome is said to be inherited by an autosomal recessive pattern ; however Mc Farlane (1947) and Latta *et al.* (1971) reported some cases with an autosomal dominant trait. Phenotypical differences of the dominant and recessive types have not been delineated, although Hall (1975) suggested that the „flat face" is less striking and abnormalities such as syndactyly, cleft palate, genital anomalies, and severe short stature are more frequent in the recessive form (5).

Our patient's trait seems to be sporadic because of the absence of family history and the absence of features which Hall already described. Autosomal recessive inheritance is also possible since his parents are first cousins.

REFERENCES

1. Habermann E. T., Sterling A., Dennis R. I. Larsen's syndrome : A heritable disorder. J. Bone Joint Surg., 1976, 58-A, 588-561.
2. Hall J. G. Arthrogryposes (Multiple congenital contractures). In : Emery A. E. H., Rimoin D. L., eds. Principles and practice of medical genetics, Churchill Livingstone, Edinburgh, London, Melbourne, New York, 1990, p. 1006.
3. Jones K. L. Smith's recognizable patterns of human malformation, W. B. Saunders Company, Philadelphia, London, Toronto, Montreal, Sydney, Tokyo, 1988, 246-247.
4. Larsen L. J., Schottstaedt E. R., Bost F. C. Multiple congenital dislocations associated with characteristic facial abnormality. J. Pediatr., 1950, 37, 574-581.
5. Maack R. W., Muntz H. R. Ossicular abnormality in Larsen's syndrome : A case report. Am. J. Otolaryngol., 1991, 12, 51-53.
6. McKusick V. A. Mendelian inheritance in men, Vols. I-II. Baltimore, London : Johns Hopkins University Press, 1992, p. 651, p. 1497.
7. Micheli L. J., Hall J. E., Watts H. G. Spinal instability in Larsen's syndrome. J. Bone Joint Surg., 1976, 58-A, 562-565.
8. Stanley C. S., Thelin J. W., Miles J. H. Mixed hearing loss in Larsen's syndrome. Clin. Genet., 1988, 33, 395-398.
9. Steel H. H., Kohl E. J. Multiple congenital dislocations associated with other skeletal anomalies (Larsen's syndrome) in three siblings. J. Bone Joint Surg., 1972, 54-A, 75-82.

SAMENVATTING

E. F. PERÇÍN, S. PERÇÍN, I. SEZGIN, A. K. AKBAS. Syndroom van Larsen met doofheid van het gemengde type.

Doofheid van het gemengde type is uiterst zeldzaam in het syndroom van Larsen, gekenmerkt door multipele luxaties en een typische facies. Eén geval van syndroom van Larsen met bilaterale doofheid van het gemengde type wordt gerapporteerd.

RÉSUMÉ

E. F. PERÇÍN, S. PERÇÍN, I. SEZGIN, A. K. AKBAS. Le syndrome de Larsen avec surdité de type mixte.

La surdité de type mixte est extrêmement rare dans le syndrome de Larsen, caractérisé par des luxations multiples et un facies typique. Les auteurs présentent un syndrome de Larsen avec surdité bilatérale de type mixte.