Clinical records

The oto-palato-digital syndrome

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The oto-palato-digital syndrome was first described by Taybi (1962) as 'generalized skeletal dysplasia with multiple anomalies'. Since then more cases have been reported all figuring patients with a complex of symptoms including deafness, dwarfism, cleft palate, characteristic facies and generalized bone dysplasia (Langer, 1967; Dudding *et al.*, 1967; Jäger and Refir, 1968; Aaie, 1969; Turner, 1970).

Only 12 cases have been reported in the available literature. This paper reports a further case of this condition and an unsuccessful attempt to improve hearing by stapedectomy.

Report of a case

A ten-year-old boy was admitted to the Rothschild University Hospital for surgical treatment of right undescended testis.

He was born following an uneventful full term pregnancy by assisted breech delivery with footling presentation. His birth weight was 3,750 gr., length 55 cm. and head circumference 36 cm. He walked at 18 months, and spoke at 20 months. At school his progress was slightly below average and his speech unclear.

The parents are cousins of the second degree, his six brothers and sisters had no physical abnormalities.

Physical examination

The patient was short and slim, 132 cm. height and 26·5 kg. weight (below the 10th percentile for age). The head had a circumference of 56 cm. (98th percentile for age), and had a prominent occiput. His face showed frontal bossing, hypertelorism, antimongolian slant, and a broad nasal bridge (Figs. 1, 2).

The teeth were irregular, with gingival hypertrophy (Fig. 1). The palate showed a very high arch, but no cleft was evident. The external ears were slightly low set but normally shaped (Fig. 2). The ear drums were normal.

The torso was small and funnel shaped. Both hands showed digits with broad distal phalanges, the thumbs long and deviated laterally. The second digits of the feet were of increased length (Fig. 3). The right testis was palpable in the right inguinal canal. Chromosome lymphocyte studies were normal.

Figure 4 shows the audiological findings (severe bilateral conductive deafness).

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FIG. 2.
The same patient in profile, the external ears slightly low-set, but normally shaped.



FIG. 1.
Shows frontal bossing, hypertelorism, a broad nasal bridge, irregular teeth with gingival hypertrophy.

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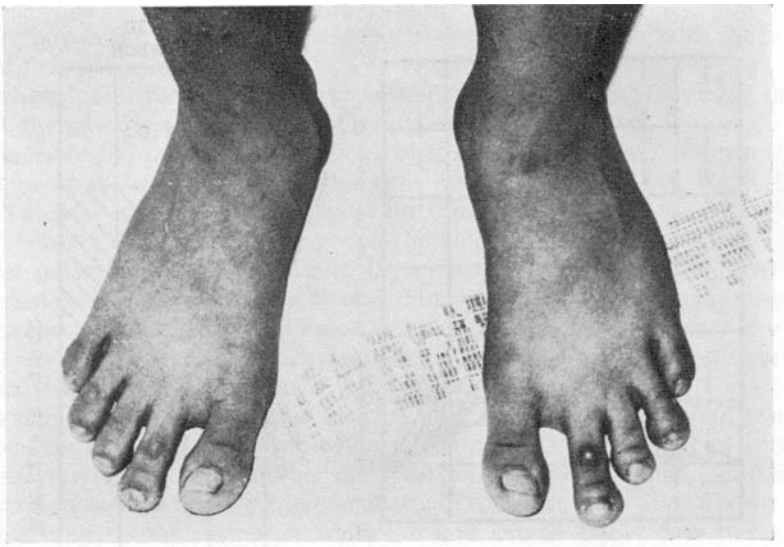


FIG. 3.
Second digits of the feet of increased length.

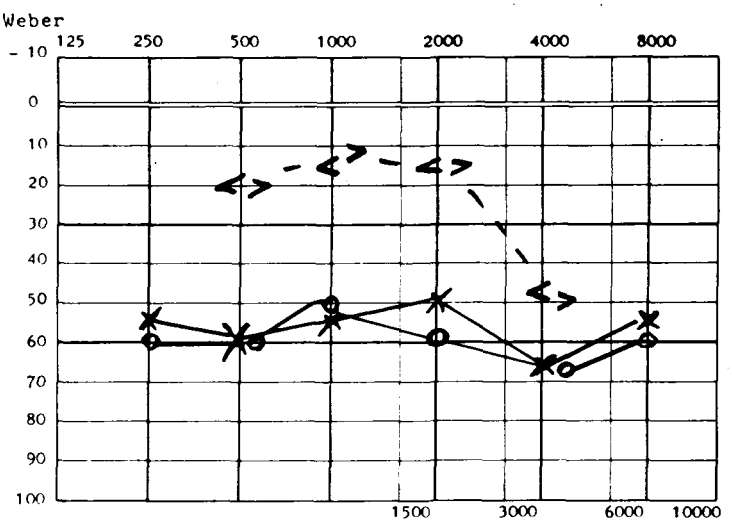


FIG. 4.
The audiological findings—severe bilateral conductive deafness.

Figure 5 shows the tympanometric findings—low compliance, absent stapedius reflex and a characteristic curve of poor motility of the ossicular chain.

Radiology of the skull, mastoids and pyramids showed metopic suture, absent frontal sinuses, poor pneumatization of the mastoid and thick ossicles. Lumbar spina bifida and open sacral canal were also noted.

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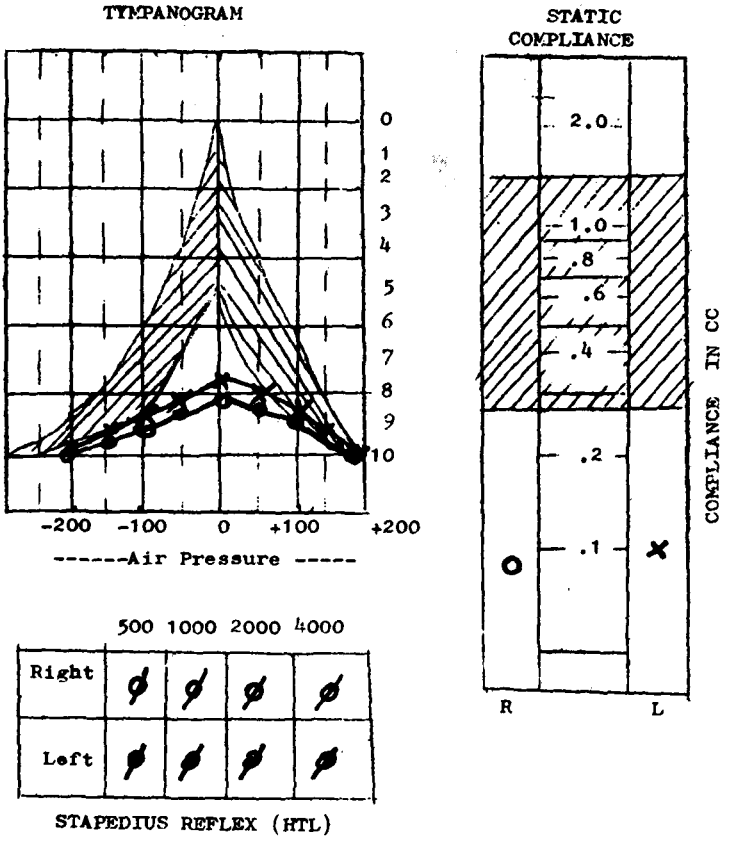


FIG. 5.

Shows the tympanometric findings—low compliance, absent stapedius reflex and a characteristic curve of poor motility of the ossicular chain.

Clinical course

Right exploratory tympanotomy was performed, the long process of the incus was found to be thickened and the crura of the stapes widened. The malleus appeared normal. There was no motility of the ossicular chain. The bony covering of the facial nerve appeared to be very thin. Stapedectomy was performed and a wire prosthesis was placed in the oval window and fixed on the long processus of the incus. The round and oval windows had a normal appearance. Good motility of the ossicular chain appeared to follow the introduction of the prosthesis.

Six days after operation right peripheral facial nerve paresis appeared. The paresis improved gradually and full recovery was attained six weeks later.

The ear remained dry but there was no hearing improvement after the operation. Left tympanotomy was not performed because of the refusal of the parents.

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Discussion

This boy showed the common features of this rare syndrome, with the exception of the cleft palate.

Although first thought to be transmitted as an autosomal dominant or sex linked, the actual genetic inheritance is not yet determined.

Langer (1967) presented three boys of the same family and Turner (1970) found two half brothers having the same syndrome. Jäger and Refir (1968) as well as Aare (1969) reported cases of the syndrome in female patients. Gorlin (1970) believes that there may be genetic heterogeneity in this condition.

The patient described by Taybi (1962) had conductive hearing loss and thickened ossicles as showed by X-rays, but no tympanotomy was performed. Buran and Duvall (1967) performed tympanotomy in two patients, in which were found abnormal ossicles. Stapes mobilization and stapedectomy were performed, without success, in an attempt to improve hearing.

In our case we found coarse ossicles with reduced mobility. We tried to improve the hearing by performing stapedectomy. Although good motility of the ossicles was established during the operation, audiological findings after the operation showed no hearing improvement.

This supports the suggestion of Buran and Duvall (1967) that the course ossicles do not transmit vibrations normally.

Summary

A boy aged ten years with oto-palato-digital syndrome is discussed.

Because of severe conductive hearing loss tympanotomy was performed and abnormal poorly mobile ossicles were found.

Stapedectomy was performed without improvement of hearing.

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