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Duplication of the pinna (polyotia) in a case of Brachmann–de Lange syndrome

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Summary—A rare case of polyotia in Brachmann–de Lange syndrome (Goodman and Gorlin, 1977) is presented and a method for the surgical correction of the duplicated auricle briefly outlined. A review of relevant literature is included in the discussion.

Case report

A 5-year-old female child, M.I., was brought by her mother for treatment of an additional right ear. The child was born after a full-term normal delivery of consanguineous parents. There was a history of a failure to thrive, bouts of asphyxia and a hoarse cry soon after birth. The child's physical growth was stunted and speech was limited to the use of words such as "ma-ma, da-da", *etc.* Her appearance is shown in Fig. 1.

Local examination showed a normal left ear except for a single accessory auricular tag in front of and above the tragus. The right ear showed a normal pinna and external auditory canal but the tragus was rudimentary. The tympanic membrane was normal and mobile. A smaller auricle which was a mirror image of the former was present. It was fused to the normal auricle from the incisura between the helix and tragus to the intertragic incisure, and contained cartilage. This accessory auricle showed a rudimentary concha and external auditory canal which ended in a skin-lined cul-de-sac, lateral to the temporo-mandibular joint. A thimble-like extension of cartilage continuous with that of the duplicated auricle intervened between this cul-de-sac and the joint. Below the duplicated auricle and separate from it was a tag of fat covered by skin which might well have been a duplicated lobule (Fig. 2).

The child's physical activity was sluggish. On general examination the height was 84 cm and she weighed 10 kg. The head circumference measured 44 cm. An IQ test showed a quotient of 55. The hearing was apparently normal. Roentgenograms revealed spurs at the anterior angle of a micrognathic mandible. The heads of the mandible were flat and placed in a shallow glenoid fossa. Schuller's projection (Shambaugh, 1967) showed absent pneumatisation of both mastoids except for well developed mastoid antra.

Surgical correction

Surgery was performed under endotracheal general anaesthesia. An incision was made between the normal and additional pinna, to circumscribe the additional lobule. Surgical effort consisted of excision of this additional lobule and trimming of the excess cartilage and skin of the duplicated auricle. An adequate amount of tissue was left behind to reconstruct the tragus (Fig. 3). A pleasing result was obtained (Fig. 4).

Discussion

The de Lange syndrome or "Amsterdam type of mental defect with congenital malformations" was first described by Brachmann in 1916 and its features were expanded upon by Cornelia de Lange in 1933. Hence this syndrome is known as the Brachmann–de Lange syndrome (Goodman and Gorlin, 1977). Its incidence is estimated at approximately 1:10,000 live births (Schuster and Johnson, 1966) and is characterised by mental retardation in association with a cluster of minor congenital malformations. The peculiar facial features make it possible to recognise the condition on inspection (Jervis and Stimson, 1963). Among the physical malformations are short stature, eyebrows which meet in the midline (synophrys), long eyelashes, hypertonicity of the muscles and sluggish physical activity. True hypertrophy of the muscles has also been reported (Barve *et al.*, 1984). The lips are thin. A midline beak in the upper lip corresponds to a notch in the lower lip and the angles of the mouth curve downwards. The nostrils are anteverted. Other features described include small hands with proximally implanted thumbs, and a simian crease. Lanugo-like hair covers the face and body; also described are microbrachycephaly and a micrognathia with spurs at the anterior angle of the mandible (Smith, 1970). A number of orthopaedic anomalies can occur (Joubin *et al.*, 1982). Our patient showed all the above features and, in addition, showed a polyotia of the right ear.

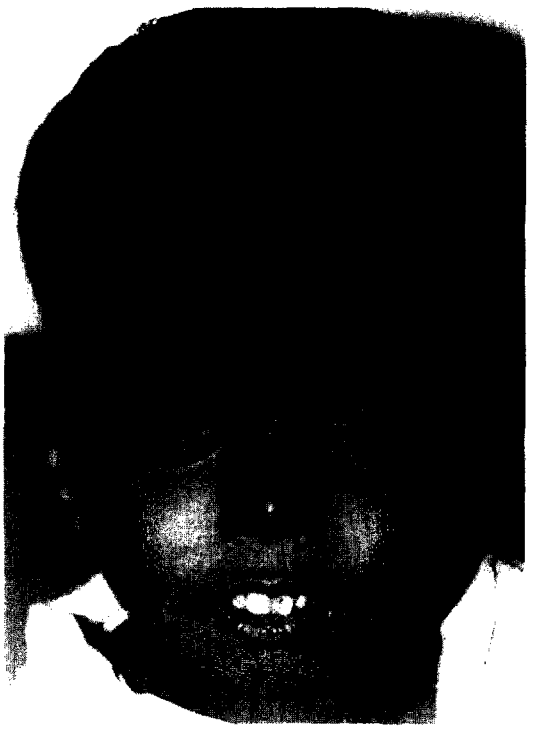

Fig. 1

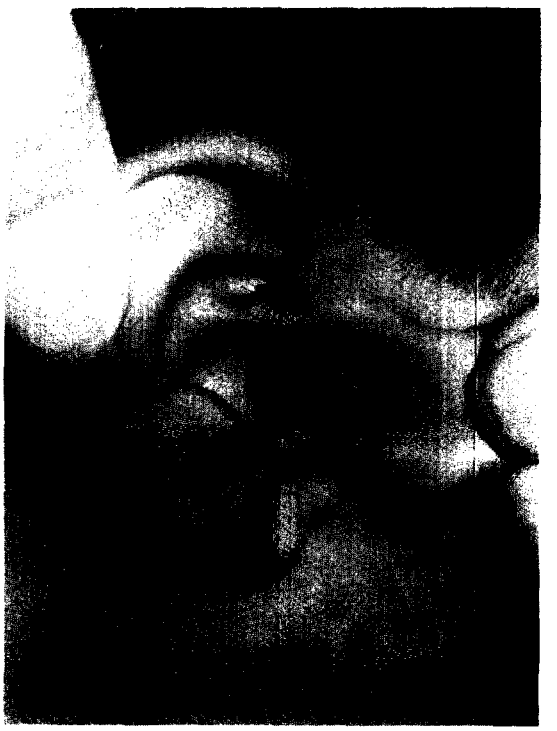

Fig. 2

Fig. 3

Fig. 4

Figure 1—Facial view of patient. Figure 2—Appearance of right ear. Figure 3—Intra-operative view of excision of accessory ear showing abnormal cartilage. Figure 4—Postoperative appearance.

Among the deformities of the external ear polyotia is extremely rare (Birrell, 1978). The patient described by Bol and de Kleyn in 1918 had a hare lip on the left side with a cleft of the hard and soft palate. He also had a dermoid cyst on the left side at the edge and somewhat under the left temporal cornea. The right ear had two auricular tags and the left ear had two auricles growing as mirror images of each other. Both were fully developed but a normal tragus was lacking. In our case the additional auricle was slightly smaller than the normal one. Though the photograph in their article does show an infant with a low hairline, a micrognathia and a mouth with downward turning of the angles, no further description is given and we will perhaps never know if their patient had any features of the Brachmann–de Lange syndrome with polyotia. Low-set ears along with choanal atresia have been classified as occasionally occurring defects in this syndrome (Smith, 1970).

The aetiology of this condition is uncertain but it may be due to single gene mutations. Chromosomal aberrations of different types have occasionally been found but they have not been consistent enough to be considered a mode of determination (Smith, 1970). An autosomal recessive inheritance has also been postulated (Optiz *et al.*, 1964). In our search of the literature we were unable to find another case with a similar ear defect in this syndrome.

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