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Familial Microtia with Meatal Atresia in Father and Son*

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Microtia is a rare congenital ear malformation which is characterized by gross hypoplasia or aplasia of the pinna of the ear, with a blind or absent external auditory meatus.

Congenital microtia associated with absence of external auditory meatus is a common cardinal finding of some syndromes such as The First Arch and the Treacher Collins Syndrome, but familial microtia and auditory atresia are rarely reported in medical literature. However, Ellwood et al[1] observed unilateral microtia with meatal atresia in 2 siblings whose parents did not show the trait. Genetic transmission of such malformations has not been described previously.

The purpose of this paper is to report a father and son, who both had microtia with meatal atresia. This suggested that the transmission pattern of microtia might be dominant.

Case Reports

This was a two-year-old male who was suffering from congenital deformity of the right ear. He was the sixth child of unrelated parents. The father had the same ear malformation on the same side. The family history was negative for similar anomalies. All other siblings in the family had normal ears.

There was no history of radiation exposure, drug ingestion or febrile illness during the pregnancy. The child was found to have a very small and deformed right auricle, and absence of external auditory meatus on the same side. At the usual site of the ear, there was only a soft tissue

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mass beneath the skin, raised above the surrounding tissue plane. The left ear was normal.

The right microtia and external auditory meatal atresia were also found in the father (Figures 1 and 2). There was no other congenital or structural anomalies in either father or son.

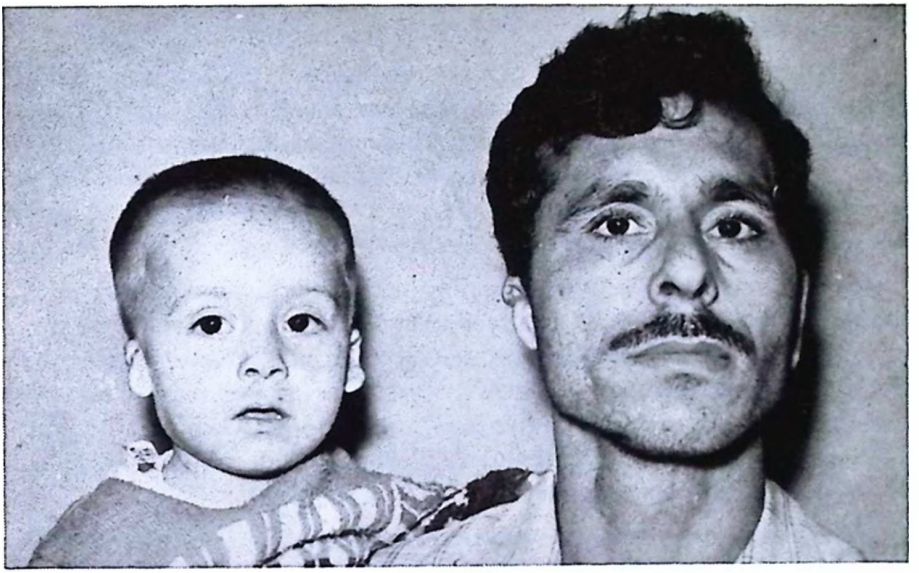

Figure 1

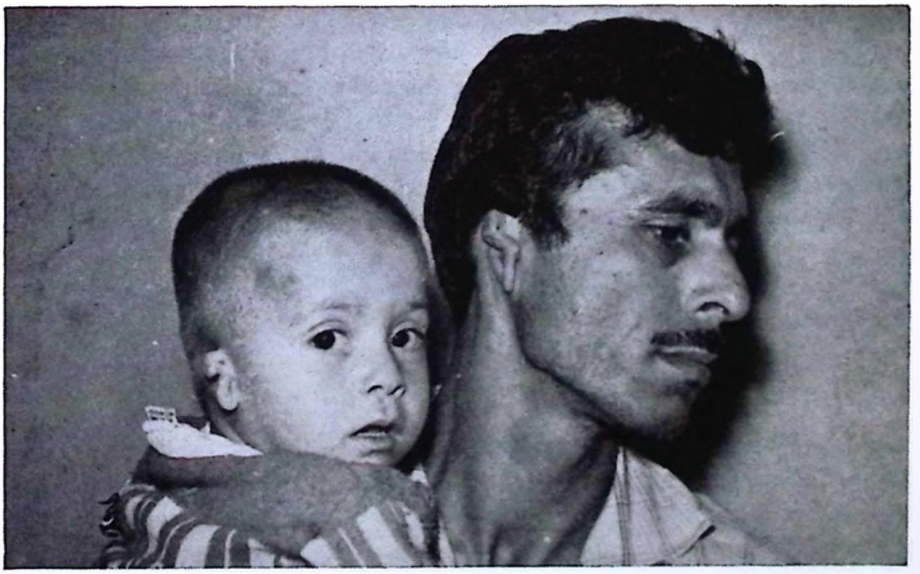

Figure 2

Radiological examinations of the father and son included normal chest films, a normal intravenous pyelogram and skull films indicating that the bony structure of the inner ear was structurally complete. Chromosomal analyses were performed from lymphocyte cultures of father and son and these showed normal male karyotypes (46, XY).

Discussion

Congenital microtia is a rare congenital ear malformation. This malformation is associated with some syndromes such as, First Arch, Treacher-Collins syndrome and some hereditary ear malformations.[2, 3] In addition, bilateral microtia or anotia with atresia of the external meatus was observed by Livingstone,[4] in approximately 15 % of children with halidomide embryopathies.

This more serious form of microtia usually is unilateral; in 31 cases reported by Hanhart,[5] only 6 were bilateral. Whetnall and Fry[6] reported that microtia is more common in the right ear, as was found in our cases. Asymmetry of the face is usually associated with unilateral microtia and meatal atresia,[7] but was not present in our family. Auricular fistulas or appendages, cleft or high palate, and middle ear disease usually complicate the disorder.

The mode of transmission of microtia or anotia has not been established previously in medical literature. Minimal hereditary deformations of the auricles, such as marginal ear pits, pre-auricular appendages, or flapped ears have been reported by Fourman and Fourman[2] and Wildervanck,[3] in members of families having microtia with meatal atresia. These reports suggested the dominant inheritance of microtia and meatal atresia. Ellwood et al[1] observed unilateral microtia with meatal atresia in 2 siblings whose parents did not show the trait. The absence of anomalies involving the ears in previous generations in both families, and the known consanguinity in one of the family's, suggested recessive factors. In our family, the father and the son showed the same malformations on the same side. This might be evidence that transmission of microtia is possibly dominant.

Summary

Familial microtia with meatal atresia without any other congenital malformation is uncommon. A father and son with microtia and meatal atresia are presented; father to son transmission has not been reported previously. This report gives a speculative suggestion on the mode of inheritance of microtia.

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