Familial Microtia with Meatal Atresia in Two Sibships*

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Hereditary malformations of the external ear, such as microtia and auditory meatal atresia, unassociated with other congenital defects or syndromes, are rarely reported in the medical literature. McKenzie (1958) described such deformities of the external ear as one of the phenotypic variations within the category of a 'first arch syndrome' caused by a dominant gene with variable penetrance. Complete penetrance of the first arch syndrome gene is proposed to result in the clinical entities known as Treacher Collins syndrome, Pierre Robin syndrome, or mandibular dystosis (McKenzie, 1966). A recent paper (Peterson and Schimke, 1968) challenged this concept and concluded that there were several genetically distinct and simply inherited branchial arch syndromes which had phenotypic overlap. Fraser (1964) introduced the probability that only a proportion of cases with a first arch syndrome defect were genetic and that many unilateral cases were phenocopies.

The physician who must provide genetic counselling to parents of an infant with malformations of the external ear not associated with other defects and with a negative family history could be understandably optimistic after a review of available literature. The purpose of this paper is to report two sibships, in each of which malformations of the external ear occurred in two successive pregnancies.

Case Reports

Family A

Case 1. This male, first child of Arab Moslem first cousin parents, related through their fathers, was the product of a full-term pregnancy, complicated only by mild pre-eclampsia, and of a normal delivery. There was no drug ingestion, radiation exposure, or illness during the pregnancy. Birthweight was 2700 g. The child was found to have bilateral absence of the auricles (anotia) and external meatal atresia with the external

meati represented only by small dimples. At the usual site of an ear, there was only a soft tissue mass beneath the skin raised above the surrounding tissue plane; there were no cartilage remnants. His jaws were described as difficult to open at birth, and difficulty in swallowing, with regurgitation, was noted when feeding began. There was slight hypertelorism of the eyes, but the face otherwise appeared normal. The lungs, heart, abdomen, genitalia, and extremities were without structural anomalies; the pupils reacted normally to light stimuli and tendon reflexes were intact. On the seventh day of life, in the absence of fever, the infant had a single generalized motor seizure, the aetiology of which was not determined. Diagnostic radiological examinations included a normal chest film, a barium swallow within the normal limits, a normal intravenous pyelogram, and routine skull films indicating that the bony structure of the inner ear was structurally complete. In audiometric testing at 1 week, noises louder than 80 decibels elicited a startle reaction. The child was readmitted to the hospital at 2 months with pneumonia. Difficulty with swallowing was again noted and oesophageal stricture was suspected but not proven. He was discharged after several weeks, but died at home at 4 months of age. Necropsy was not performed.

Case 2. The second offspring of this union was a female, the product of a pregnancy without complications, and of a normal delivery. Birthweight was 2760 g. Bilateral absence of the auricles and external auditory meatal atresia were also found in this child. The ears were represented only by soft tissue beneath the skin. Facial structures were without gross abnormality, but over-all appearance was described as 'strange'. Lungs, heart, abdomen, genitalia, and extremities were normal. Guthrie blood test on second day showed a slight increase in blood phenylalanine, in the range of 2 to 4 mg./100 ml.; a repeat on a blood sample taken at 2 months was normal. Chromosome analysis revealed no abnormalities. At 4 months, audiometry was attempted, but strong sound stimuli next to the head elicited no reaction. No other diagnostic tests were performed and the structural status of the inner ears was unknown. Six months after birth, the mother reported the death of the child at home. The cause of death was unknown, but was probably infectious illness with lack of medical attention. Necropsy was not performed.

The family history for similar anomalies is negative. A third child, a girl, has normal external ears. It

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should be noted that first cousin marriages are extremely common (over 25%) in Arab unions.

Family B.

Case 1. This male child, first offspring of a Jewish marriage, was the 3650 g. product of an uncomplicated pregnancy and term delivery. At birth, microtia and external auditory meatal atresia on the left side were found. The auricle, located in normal position, consisted of a continuous ridge of cartilage covered by skin and appeared as if the auricle had been folded forward. There was a normally formed ear lobe. The right auricle showed only partial downward folding of the upper pole of the helix; there was atresia of the right meatus. Facial structures appeared normal, and asymmetry was not present. The tongue had a short frenulum. Lungs, heart, abdomen, genitalia, and extremities were without anomalies. At the age of 18 months, the child had an operation for tongue-tie, and an adenoidectomy. Growth and psychomotor development have been within normal limits. Audiometric testing showed a hearing loss of 70 decibels by air conduction and 30 to 50 decibels by bone conduction. A bone conduction hearing aid is now used at the left ear. The status of middle ear structures is unknown since attempted diagnostic procedures have been unsuccessful. Surgical construction of the external auditory canal and tympanic membrane has not been attempted. Now aged 4 years, the child has considerably retarded speech development secondary to hearing loss.

Case 2. The second child was also a male; pregnancy and delivery at term were uncomplicated. Birthweight was 3000 g. On examination, microtia and external auditory meatal atresia of the left ear were found. The structure of the anomalous auricle was identical to that of the older sib; there was also a similar folding of the upper helix on the right. The right ear was otherwise normal. The face was without structural anomalies or asymmetry. A small pilonidal sinus with 1 cm. depth was present. Lungs, heart, abdomen, genitalia, and extremities were normal. Transient hyperbilirubinaemia without anaemia or evidence of haemolysis was present in the neonatal period. Chromosomes were normal. Audiometric testing shows that the child is not deaf. At age 20 months, he is within normal range for growth and psychomotor development.

There is no family history for congenital anomalies of the ears or face. The parents deny consanguinity. The paternal grandparents are Ashkenazi, from Rumania. The maternal grandfather is Ashkenazi from Rumania but from a different geographical region, and the maternal grandmother is Sephardic from Greece. The mother has no sibs, but the father has a brother with two normal offsprings and an unmarried sister.

Discussion

The defects in the sibships presented in this report, bilateral anotia with meatal atresia in family A and unilateral microtia with meatal atresia in

family B, have not completely followed the pattern of auricle malformation with meatal atresia usually described in the literature. Congenital meatal atresia is usually only found associated with severe malformation of the auricle, is more often unilateral than bilateral, and is more common in the right ear (Whetnall and Fry, 1964). The anomaly in family A was bilateral, and the microtia in family B was on the left side. Asymmetry of the face, usually associated with unilateral microtia and meatal atresia (Holmes, 1949) was not present in family B. Except for minimal hypertelorism in one child of family A, there were no other structural defects such as found in the first arch syndrome or described in families with hereditary malformations of the ears (McKenzie, 1958; Fourman and Fourman, 1955; Wildervanck, 1962).

The apparent difference in the degree of hearing impairment in the two children in family B may be explained by the assumption that the hearing defect is secondary to the involvement of the ossicles within the middle ear and is not a defect of the inner ear or neural tract. Some degree of deafness, usually conductive, exists in all cases of meatal atresia (Wildervanck, 1962), but since the outer and middle ears have different embryological development from the inner ear, the neural structures of the inner ear can be normal in function (Livingstone, 1964).

It may be presumed that the anomaly in each sibship is familial, without being able to assign a dominant or recessive gene as responsible. The defects were different in the two families; and therefore their aetiological similarity is unknown. In family B it may be assumed that the expressivity of any responsible genetic factor was different within the sibship. Similarly the same defect occurred in both male and female in family A, thus excluding sex variation in expression of this defect.

The importance of genetic aetiology or the mode of inheritance of microtia or anotia with external meatal atresia has not been clearly delineated in the literature. The defects may be due to a dominant gene or genes with variable expressivity and penetrance, which control the embryological development of the stapedia artery and cause the defects grouped in the first arch syndrome described by McKenzie (1966). Dominantly hereditary cases have been reported in which microtia with meatal atresia has occurred in members of families having hereditary minimal deformations of the auricles, marginal ear pits, pre-auricular appendages, or flapped ears (Fourman and Fourman, 1955; Wildervanck, 1962). Though dominant inheritance of microtia and meatal atresia is reported,

Wildervanck (1962) believes that the majority of cases are sporadic. Phenotypically similar branchial arch syndromes may be genetically distinct, with possibly different inheritance patterns (Peterson and Schimke, 1968). With such variation of opinion about the aetiology of these defects, it is difficult to provide genetic counselling that includes the risk of recurrence in cases where family history is non-contributory.

The literature on similar defects and the occurrence in two successive pregnancies in each sibship in this report favour dominant inheritance of these two entities. However, the absence of anomalies involving the ears in previous generations in both families and the known consanguinity in family A suggest recessive factors.

Additional reports on familial cases and sporadic cases are needed to provide data from which a risk factor for recurrence of severe malformations of the external ear can be determined.

Summary

Two sibships with microtia and meatal atresia are described. In one family, two children had bilateral anotia with meatal atresia. The second family had two boys, one with unilateral microtia

and bilateral meatal atresia and the second with unilateral microtia and meatal atresia.

Familial microtia with meatal atresia, unassociated with other congenital anomalies, in sibs with no other family history, is uncommon. Opinions on the mode of inheritance vary, and genetic counselling on the risk of recurrence is presently difficult.

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