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Inheritance of an Unusual Ear Type in Man

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SUMMARY

An unusual ear type in man involving a folding of the helix is described in a pedigree with three generations. The trait seems to be due to an autosomal dominant gene with variable expressivity.

In an attempt to study their mode of inheritance Gates (1948) has compiled various types of ear shapes. (For an illustration of a normal ear, cf. Fig. 2). One of the unusual types has a folding of a part of the helix so that it touches the anthelix. This was originally reported in a mother and a child by Haike (1928). A survey of literature by the authors failed to reveal any other pedigree with this rare trait. Recently, we

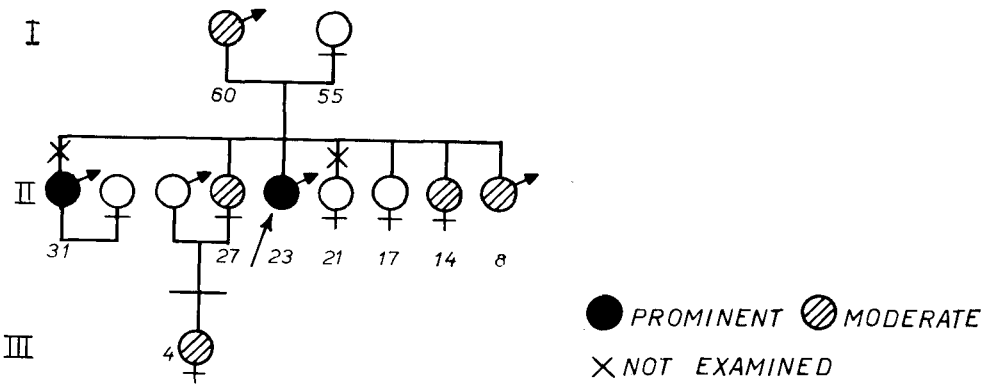

Fig. 1. Pedigree of the unusual ear type

have come across a Punjabi family in which the trait runs in three generations affecting seven individuals (Fig. 1). In each of the affected individuals, the two ears are very similar. To the best of our knowledge the trait does not seem to be associated with any hearing defect. Hence, it does not seem to have any clinical value. However, it is important from the genetic point of view and may be of significance in anthropological studies.

An analysis of the pedigree under our study (Fig. 1) shows that: (1) the trait is transmitted directly from affected person to affected person without skips or breaks in continuity; (2) the two sexes are affected approximately in equal numbers; and

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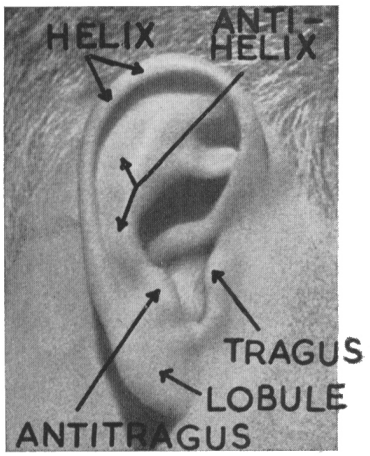

Fig. 2. Normal ear

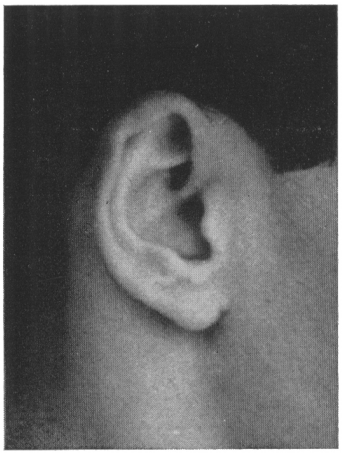

Fig. 3. The propositus (II-3) showing a prominent folding of the helix so that it almost touches the anthelix

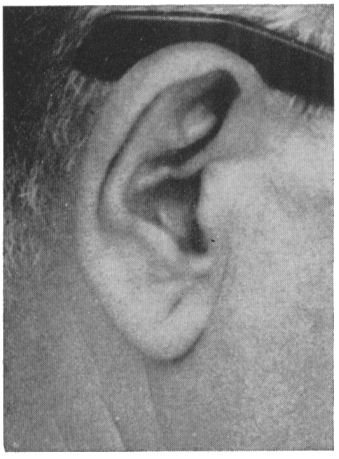

Fig. 4. Father of the propositus (I-1) showing a moderate folding of the helix

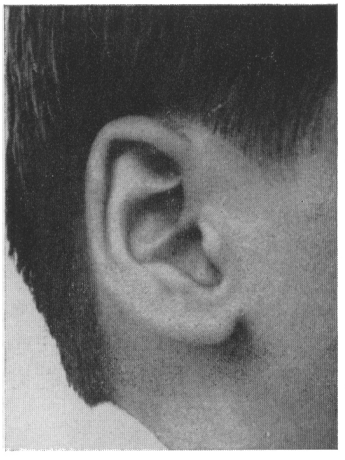

Fig. 5. Brother of the propositus (II-7) showing a moderate folding of the helix

(3) some of the children of the affected individual show the trait whereas others do not. From these observations it may be concluded that the trait is due to a single dominant rare autosomal gene.

Not all the affected members of this family have an equal expression of the trait. In some the trait is prominent, i.e., the helix is folded and it almost touches the anthelix; this gives the ear margin a notchy appearance (Fig. 3). In others it is only par-

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tially expressed, i.e., the helix is slightly folded and it does not touch the anthelix; in other words, the notch is not prominent (Figs. 4 and 5). Hence, the gene controlling this peculiar trait has a variable expressivity, perhaps due to modifiers.

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RIASSUNTO

Viene descritto un tipo insolito di orecchio, con piega dell'helix, in una genealogia con tre generazioni. Tale carattere sembra dovuto ad un gene autosomico dominante ad espressività variabile.

RÉSUMÉ

On décrit un type inhabituel d'oreille entraînant un pliage du hélix dans une généalogie comprenant trois générations. Le trait semble être dû à un gène dominant autosomique avec expressivité variable.

ZUSAMMENFASSUNG

Eine ungewöhnliche Typ des Ohres beim Menschen, die eine Faltung der Ohrleiste weist, ist in einem Stammbaum mit drei Generationen beschrieben. Die Erbeigenschaft ist offenbar durch ein autosomales dominierendes Gen variabeler Expressivität bedingt.

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