The genetics of deafness

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D. THE GENETICS OF DEAFNESS

Laboratory detection of heterozygotes of deafness. A Békéshy-audiometric examination of parents with children deaf from birth

Two types of individual transmit hereditary diseases to their children: those who have the disease in manifest form and those who without apparent disease themselves bear genes for the disease.

About 5 per cent of the children in our Swedish schools for the deaf are born of totally deaf parents or of parents with impaired hearing. The parents of the remaining 95 per cent consider their hearing to be normal.

According to the mendelian laws, a dominant character manifests itself even if the gene in question is inherited from only one of the parents. If the character is recessive, it is necessary that a gene for the disease be inherited from both parents.

According to modern opinion, the concept of dominant and recessive characters has a certain value from the pedagogic point of view, but from the genetic viewpoint a disease is neither wholly dominantly nor wholly recessively transmitted but rather relatively transmitted, depending upon the ease with which the tendency can be demonstrated in heterozygotes. In certain diseases the mode of inheritance is intermediate. If an individual is heterozygous for a disease, he has one normal and one mutant genes deviate chemically from those of normal genes. The individual may be symptom free, but, if he is subjected to sufficiently delicate physical or chemical tests, it can be demonstrated through deviations from the normal that he is a bearer of a disease factor. Such genetic carriers have been discovered in a number of blood diseases and metabolic disorders.

In an effort to solve the problem of detecting suspected bearers of a hereditary deafness factor, probably with small deviations from the normal, the writer examined, by means of Békéshy-audiometry, parents with so-called normal hearing whose marriage has produced offspring with impaired hearing probably dating from birth. The material consisted of parents in 41 families and their 112 children.

Analysis of the parent's audiograms revealed considerably more frequent and more extensive auditory impairments than are usual in the
population as a whole. It is surprising to observe that usually only one of the parents has impaired hearing. Only unusual and extensive loss of hearing with least octave range was considered. The hearing loss in the children was of three different types. That these types are hereditary is evident from the fact that children of parents deaf from birth present the same types. A further observation that would seem to support the hypothesis of hereditary origin is the similarity between the hearing impairments caused by rubella virus during the first three months of pregnancy and these assumed hereditary impairments. The rubella damage is exogenic in origin, to be sure, but the similarity between a mutant gene and a virus with respect to chemical composition and mode of action is great since both virus and genes are composed of protein bound to nucleic acid. Owing to its nucleic acid content, the genes control the individual cell and the differentiation of organs and tissues through their "steering" of the enzymes. When there is a change in the composition of the normal nucleic acid, so-called mutation, an abnormal "steering" of the enzyme that is responsible for the development of the acoustic system arises, and auditory impairment results. This is also true in the case of virus. Since the cells stop synthesizing their own nucleic acid when attacked by the virus and instead produce virus nucleic acid, there is an abnormal enzyme development with resultant hearing damage. The mutant gene for auditory impairment as it appeared in the majority of the cases investigated seems to be autosomal and dominant with variable penetrance and expressivity.

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