A GENETIC STUDY ON DEAF TWINS

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A clinical and genetic study on 25 twin pairs with at least one deaf member was performed. Concordance rate of hearing loss was 0.88 in MZ and 0.50 in DZ pairs, with an almost complete penetrance ($P = 0.94$). *Consanguinity was found in 32% of cases, corresponding to five times the average population rate in Japan.*

INTRODUCTION

The severe hearing impairment in children is a sign of many pathological conditions of auditory system caused by genetic, acquired, or unknown origin. The cases with history of hearing loss after birth are generally considered as acquired deafness. The causes of acquired deafness have been reported by many authors. It is well-known and important that maternal rubella in prenatal period, or infectious ear disease in the infant, are among the causes of deafness.

Fraser (1970) observed the identification of the cause of deafness in children. Of 2,355 deaf children 853 had unknown cause. It is often impossible to certify what is the cause of severe hearing impairment in early iife, because nobody can exclude a true genetic deafness.

In the present paper, 25 pairs of twins, at least one of whom was believed to exhibit hereditary deafness, are reported. The relative contribution of genetic factors in congenital deafness in thus measured.

METHOD

Fifthy-three pairs of twins, one or both of whom exhibited early severe deafness, were ascertained from students on lists from 30 schools for deafness in Japan. Zygosity was only determined in 32 pairs; 7 of these had a history of hearing impairments, including persistent otitis media in infant, perinatal anoxia and use of streptomycin. Twenty-five pairs of twins without a known exogenous cause of deafness were thus accepted as index cases for our genetic analysis. The zygosity was determined by similarity method, using the typing of blood groups and serum factors. The twins were considered as DZ when they were found to have at least one different blood type or serum factor.

In the first step, physical examination of each deaf subject was undertaken, including visual activity, inspection of head and neck region, and extremities.

A pair had congenital anomaly of the 5th finger of both hands with total hearing loss. However, 24 pairs had no associated systemic anomalies.

In the second step, the otologic examination was performed by senior residents at the home of children. In 4 of 32 pairs one of the twins had an evidence of chronic ear infection, which was probably a cause of severe hearing loss in early life.

In the third step, interview with parents of twins was performed, to study their family and developmental history.

By a family history, a pedigree was drawn of the parents with cousin marriage, deaf-to-deaf mating, deaf siblings or descendants. When their parents had a history of hearing loss, audiometric tests were carried out. All of the causes of hearing impairments were reviewed while studying the developmental history.

In the last step, the audiometric tests, including air and bone conduction, with or without speech reception threshold (SRT), were performed. Caloric test and rotation vestibular test were performed on only two pairs at our clinic.

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RESULTS

Thirty-two pairs with complete studies were recorded in our sample, but 7 cases had a history of acquired ear involvement. Therefore, the data of 25 pairs (17 MZ and 8 DZ) were analyzed in this report. Ten cases were teenagers. Nine cases were 20 to 24 years old, and 7 cases from 25 to 29.

Audiometric Pattern. No significant difference of hearing activities was found between the right and left ear in each zygosity group, whereas a significant difference was found between MZ and DZ twins on the same side.

Concordance and Penetrance. A twin pair was considered concordant when the difference of hearing loss between the cotwins did not reach 30 dB. Thus defined, concordance applied to 15 out of 17 MZ pairs (88%) and 4 out of 8 DZ pairs (50%). The difference is significant ($\chi^2 = 2.516$, $p = 0.10 - 0.20$). The penetrance (P) can be estimated as follows (Allen 1965): $P = 2m/(1+m) = 2 \times 0.88/(1+0.88) = 0.94$, where m is the concordant rate of MZ twins.

Consanguineous Mating. Consanguinity was found in 8 pedigrees. Its rate ($32.0 \pm 9.3\%$) was much higher than the population rate about 6% in Japan. No deaf-to-deaf mating of parents was found. In 7 of the 25 pairs, siblings or parents had a hearing impairment. In these 7 pairs, 4 were issued of a consanguineous mating.

DISCUSSION

Deafness in twins has been studied by Moos (1882), Fay (1898), Douglas (1927), Undritz (1928), Tanaka (1931), Shambaugh and Shambaugh (1933), Suzuki (1939), Lindenov (1949), Gedda (1953), and Post and Hopkins (1956). It is impossible to draw firm conclusions from these studies, for the number of cases was limited in every instance.

Thirty-seven pairs of twins with early total deafness in New York were reported by Sank and Kallmann (1963).

In their study, the concordance rate in twins estimated by audiometric study was found to be 88%

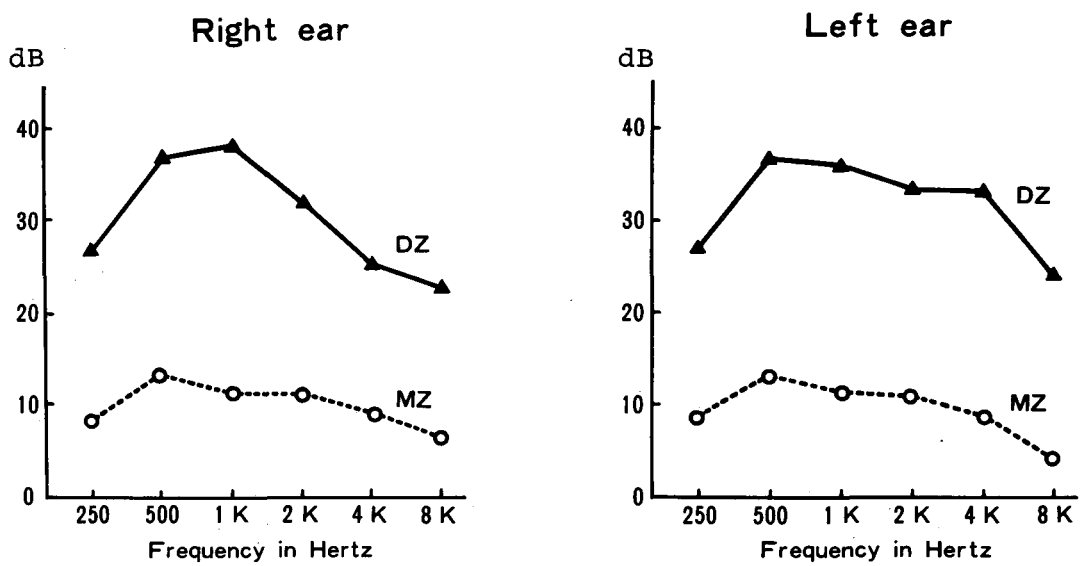

Figure. Difference of hearing activity between cotwins. The average in MZ is smaller than in DZ pairs.

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in MZ and 35% in DZ pairs. According to this analysis, the minimum number of different recessive genes for deafness has been estimated at 45 for the New York state population.

In the present study, the gene of deafness has shown an almost complete penetrance (94%). This suggests that the clinical manifestation of this gene may undergo little environmental influence.

Hereditary deafness is variously inherited, as a dominant, recessive or sex-linked (Konigsmark 1971). Autosomal recessive genes were accounted for 71.0% of congenital deafness (Furusho 1973) or for 68% (Chung et al. 1959). Except for one case of sex-linked inheritance, the present study shows an autosomal recessive model.

As the Figure shows, the average of difference in hearing loss among twins was 9 dB in MZ pairs and 30 dB in DZ pairs, and therefore a dissolution indicated a remarkable small value in MZ twins.

Heritability in degree of hearing activity was estimated as follows: on right ear, $h^2 = 0.78$; on left ear, $h^2 = 0.73$.

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