

DOMINANT CONGENITAL DEAFNESS AND PROGRESSIVE OPTIC ATROPHY

Report of a Family Through Four Generations

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Six persons in four generations had congenital severe deafness and progressive mid-life visual failure. The 53-year-old proband and her 9-year-old son, studied in detail, had severe neural hearing loss and optic atrophy, much more severe in the mother. Her vision was 20/80 in each eye while her son's vision was normal. No cause for the hearing or visual loss was present except heredity.

The proband's father, seen years ago at the age of 68, was congenitally deaf and had progressive visual loss with optic atrophy. By history, the proband's two maternal aunts and paternal grandmother had this same syndrome. The syndrome, transmitted by dominant mode, is unique and distinct from other familial syndromes of visual and hearing loss, including those described by Usher, Refsum, Alström, Cockayne, Norrie, Small, Sylvester, Rosenberg and Chutorian, and Tunbridge and Paley.

In this report we document a previously underscribed syndrome characterized by congenital hearing loss and a slowly progressive mid-life visual failure with optic atrophy, transmitted by dominant mode.

REPORT OF CASES

Case I

This 53-year-old woman was admitted to the hospital because of optic atrophy and progressive visual loss over the past four years. Four years before, she noticed mild difficulty in reading small type; this progressed slowly until at present she was only able to read fairly large type. She developed no speech in infancy; severe hearing loss was first noticed when she was about 2 years old.

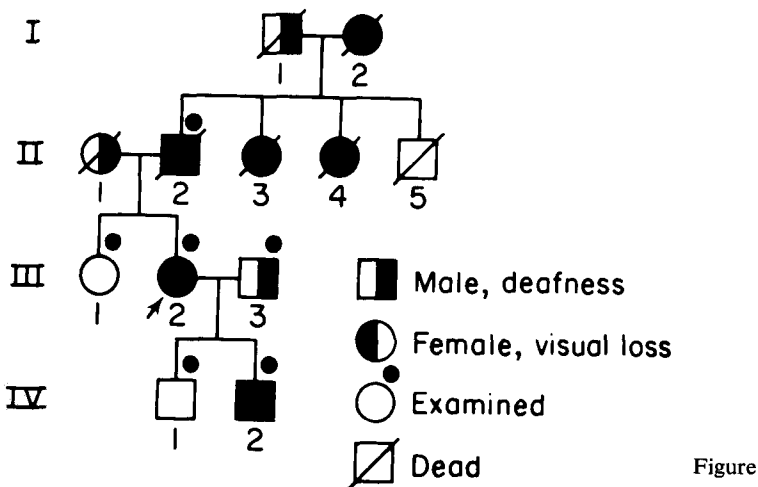
Neurological examination was normal except for deafness and optic atrophy. Ophthalmological examination found vision of 20/80 in each eye and moderate bilateral symmetrical optic atrophy. A skull series including multiple views of the sella turcica and optic foraminae was normal. A pneumoencephalogram and polytomograms of the chiasmatic region showed no evidence of a space-occupying lesion or deformity about the chiasm. A right brachial angiogram also showed no sign of intracranial vascular disease or mass. Laboratory tests and temporal bone tomograms were normal.

* Shortly after completion of this work Dr. Bruce W. Konigsmark died from leukemia, on 30 October 1973. Although this paper could not be presented to the Congress, the editor thought that it should be included in the present Congress Proceedings, where it thus represents Dr. Konigsmark's last contribution.

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Case II

This 9-year-old son of the proband was apparently normal except for severe hearing loss. The hearing loss was first noted when he was about 2 years old, although it was probably congenital for he had developed no speech. Audiometric testing at age 3 showed a moderately severe sensorineural hearing loss. At 9 years he was admitted for diagnostic studies. Examinations were normal except for profound hearing loss and mild optic atrophy. Ophthalmological examination revealed vision of 20/15 bilaterally. External and slit-lamp examinations were normal. The fundus was normal except for distinct pallor, more marked on the temporal aspect of the optic nerves.

Case III

The proband's father was born deaf and died at 70 years of age. About the age of 64 visual loss was noticed. Ophthalmological examination showed visual acuity at 20/60 in each eye and bilateral moderate optic atrophy.

Other Family Members

Three paternal aunts of the proband and her paternal grandmother were all born deaf and had deterioration of vision in their later years. They were probably affected with this syndrome.

COMMENT

Thus, a mother and her son and father had a similar syndrome of congenital deafness and progressive optic atrophy. By history, two paternal aunts and a paternal grandmother were affected. Transmission of this syndrome, involving six patients in four generations, was by dominant mode.

There are nine known hereditary syndromes in which hearing loss is combined with retinal disease. The four syndromes including hearing loss and retinitis pigmentosa, i.e., Usher, Refsum, Alstrom, and Cockayne syndromes, can be excluded because of the absence of any evidence of retinitis pigmentosa in our patients. The syndrome of sex-linked retinal malformation, mental retardation, and hearing loss (Norrie syndrome), can be ruled out because of

the absence of microphthalmia and of the characteristic retinal gliotic changes. The Small syndrome with retinal vessel changes, muscle atrophy, mental retardation, and hearing loss, is quite different from that found in our cases.

The four hereditary syndromes with hearing loss and optic atrophy are Sylvester (optic atrophy, ataxia, and progressive hearing loss), Rosenberg-Chutorian (optic atrophy, polyneuropathy, and neural hearing loss), Tunbridge-Paley syndrome (progressive optic atrophy, progressive neural hearing loss, and juvenile diabetes mellitus) and optico-cochleo-dentate degeneration. They all differ from the syndrome in our patients because of the different neurological findings. Thus, this syndrome of dominantly transmitted congenital deafness and progressive optic atrophy is unique and represents a newly described entity.