NEUROFIBROMATOSIS 1 AND CONGENITAL DEAFNESS : TWO MENDELIAN CONDITIONS SEGREGATING IN A FAMILY

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SUMMARY : A "congenitally" deaf-mute young man with autosomal dominant neufibromatosis 1 and with a deaf-mute wife is described. Deaf-mutism in the wife is due to autosomal recessive genes. The couple previously had a prematurely born infant who died immediately thereafter, and male stillborn twins. The third pregnancy is continuing under additional risk of chromosomal abnormality because of her relatively advanced age.

Key Words : *Congenital Deafness, Infertility, Neurofibromatosis-1*

Neurofibromatosis 1, NF 1, is known to be associated with a variety of conditions, congenital in nature and frequently with malignant as well as non-malignant neoplasms (Crowe et al. 1956; Friedman et al. 1982; Johnson and Charneco, 1970; Riccardi, 1981). We previously reported on associated features what we have considered of "unusual" occurrence on two occasions (Toğrul and Şaylı, 1991). We here report another case in which not only the proband himself but also his family members present still "unusual" features.

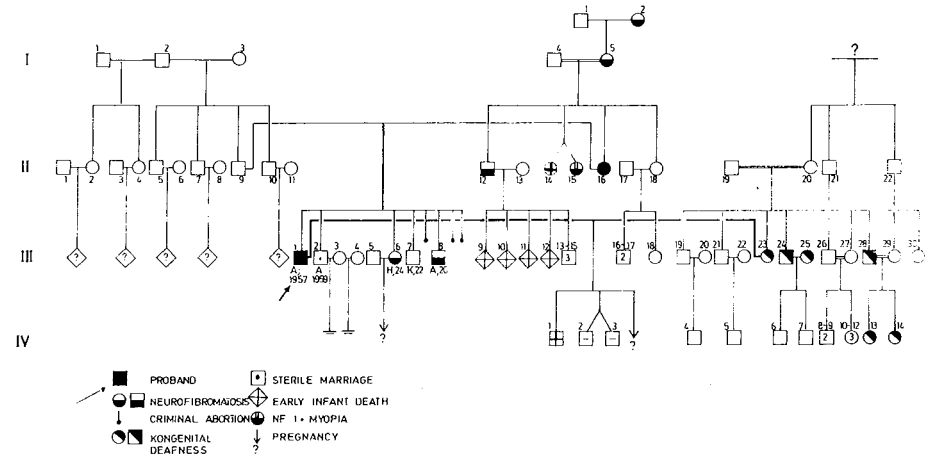

Fig. 1 : Pedigree of A. Family. Arrow indicates the proband.

tenatal diagnosis is possible.

- An upper limit of 25 percent congenital deafness-this is most intriguing and also not possible to detect prenatally. And there is an additional risk of fetal loss.

Note added in review :

She gave birth to a normal female child on 25 November 1991. The newborn seemed to be free of any of the signs and symptoms referable to the first 2 conditions.

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CORRECTIONS

(Volume 3, Number1, January 1992)

* pg. 43

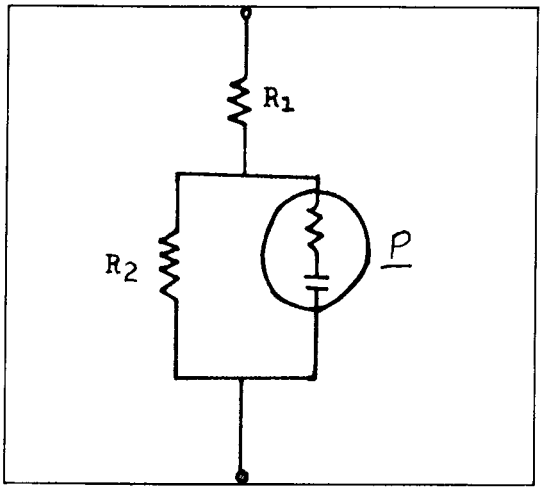
Riht column, 33. line :

"Base levels range from 10 KΩ **to 500 K**Ω cm^2, even within the same individual (Ackerman et al. 1979)."

** pg. 44 left column 26 line

" ..... was only 2 to 3 % of the **dc** value..."

*** pg 44, Fig. 2 :



**** pg. 45 Left column, 10. line :

"$C = 0.0089 \frac{KA}{t} \times 10^{-6} \mu F$ "