RECESSIVE SEX-LINKED INHERITANCE OF CONGENITAL EXTERNAL OPHTHALMOPLEGIA AND MYOPIA COINCIDENT WITH OTHER DYSPLASIAS*

A REAPPRAISAL AFTER 15 YEARS

BY

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IN 1950 a study was published (Salleras and Ortiz de Zarate, 1950) of a family with congenital external ophthalmoplegia. As this family remains the only one of its type (Klein and Franceschetti, 1964), it seemed worth while to bring the story up to date. An opportunity for reassessment after 15 years arose when a member of the family sought genetic advice.

Observations

There are now various additions to the pedigree drawn in 1950 and the sibs in each generation have been re-numbered (Figure). For example, II, 15, the only member of that sibship who lived until adulthood has children and grandchildren; III, 4, and her children have been added; etc., but there is also one fact of particular importance—III, 6 (previously regarded as unaffected) has borne an affected son.

FIGURE.—Recessive sex-linked pedigree of congenital external ophthalmoplegia.

She was not previously noted as a heterozygote because she had no children, but in the Table of dysplasias of this family (Salleras and Ortiz de Zarate, 1950, Fig. 3) it was noted (under the initials N. D.) that the tendon-jerks in the lower limbs were

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absent. This areflexia was found in all ophthalmoplegic (male) members of the family and also in all (healthy) female members who had borne affected sons; it was then considered to be a strong indication (by linkage or pleiotropy) that the affected person was a carrier. The birth of IV, 15 is a ratification of that assumption.

Only III, 3, and IV, 7, were examined personally at the present follow-up and the findings were the same as in 1950.

III, 3 (female, carrier) has areflexia, moderate enlargement of the thyroid gland, severe nervousness, sweating, and sometimes tremor of the hands. All these suspected signs of thyrotoxicosis were present in all carrier women and in no other members of the family. It could have the same indicative value as the areflexia but it is not so objective and requires a more thorough investigation.

IV, 7 (affected male) has almost complete external ophthalmoplegia, high myopia, absent tendon-jerks, anomalous occlusion of the teeth, and scoliosis (previously referred to as "thoracic malformation"). An electromyographic study of the upper limbs has revealed no abnormalities.

For the sake of completeness one thing can be added, though it surely bears no relation to the ophthalmoplegia: IV, 13 is a deaf-mute, as is her paternal grandmother.

Summary

A pedigree published in 1950, which has remained unique, is reviewed and brought up to date. The transmission is sex-linked, the female carriers being recognized by the absence of tendon-jerks.

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