Hereditary Hyaloideo-Retinal Degeneration and Palatoschisis

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Summary

The first description is of a Dutch family with hereditary autosomal dominant retinal detachment, due to degenerative anomalies of the choroid, retina and vitreous and associated with myopia varying from 7 to 18 dioptries. Pre-senile cataract is the rule.

All the patients have also palatoschisis and remarkable faces with wide flat noses and heavy protruding lower jaws. The teeth are irregular. They also have genua valga and overextension of the elbows.

Nothing is known about the eyes of three members of the family with palatoschisis who died in infancy.

The patients whom we examined showed a striking resemblance to the members of a Danish family seen by Frandsen (1966).

Discussion

Waardenburg: Are we concerned here only with palatoschisis? Or is it a forme fruste of a cheilognathopalatoschisis? In Denmark the former has been found as a separate anomaly which is often recessive, in contradiction to the irregularly dominant latter anomaly. The collection of more material for comparison is desirable in order to determine whether this is pure coincidence or whether it is really a new syndrome.

Falger is in complete agreement with this. The condition is pure palatoschisis.

Oosterhuis: You mentioned that one of your patients with retinal detachment also had malignant myopia. Was this also true for the other patients? It is possible that he have here hereditary degenerative myopia associated with hereditary palatoschisis?

Falger: The degree of myopia in the other patients varies from 7 to 10 dioptries. The possibility you suggest must not be excluded.

Jansen observes that we may not speak of Wagner's syndrome as the most important symptoms of this condition are absent.

Falger: The typical symptoms

i.e. transparent vitreous and white preretinal membrane
are usually absent

but even so Frandsen concludes that the Wagner syndrome is present in his patients.