A New Syndrome in Three Generations of a Dutch Family

J.

van den Bosch

Leiden

Summary.

A hitherto not described complex of symptoms was found in two living brothers, aged 9 and 7 years. The complex consisted of

mental deficiency;

backwards in somatic development;

horizontal nystagmus;

chorioideremia;

abnormal retinogram;

a high degree of myopia;

scapulae alatae;

an acro-keratosis verruciformis like skin abnormality;

a high susceptibility for bronchial and skin infections;

the tendency to undress and stretch out on a cold floor during hot days;

no local sweat reaction on injection of a 0.1 cc. of a solution of 1 % acetyl choline.

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The theory that a gene or a complex of genes on the X-chromosome is to be regarded as the genetic cause is discussed. The fact that only males have been or are showing the syndrome, and that the genealogical link between the sibships in which boys have been reported as affected is through the mothers and ultimately the fact that several of the symptoms of this syndrome have been described as X-chromosomal traits only seem to give evidence for this theory.

Clinically the syndrome seems best classified in the group of the so-called anhidrotic ectodermal dysplasias.

Winkelman, J.E. and Horsten, G. P. M.: Ophthalmologica 137: 423–425 (1959)

Congenital Blindness in the Presence of a Normal Fundus.

By J. E. WINKELMAN and G. P. M. HORSTEN, Amsterdam.

Entirely normal parents had a child which at the age of 6 months was presumed to be blind. We saw the boy then for the first time; he showed nystagmoid movements which sometimes developed into real nystagmus. The pupils did not react to light, there were no following movements to light, but the fundi were completely normal. The boy is now 4½ years old and the condition is unchanged. He is very intelligent. The paediatrician found one peculiarity, viz. a typical herringbone pattern of the nails.

Ten months ago a sister was born who is also blind, with normal fundi. The shape of her skull is somewhat peculiar (brachy-cephaly) with a “dip” in the occiput. She has the same nail pattern.

ERG and EEG were carried out under slight anaesthesia. The boy showed diffuse EEG disturbances, especially occipitally. In the girl there were marked EEG disorders. In both children, the ERG was absent.

Summarizing

two successive children in one family who are blind with normal fundi

absent ERG

disturbed EEG are presented. There are no further children. What kind of affection may this be?