Amaurosis congenita (Leber)

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In spite of recent publications by Waardenburg, Alström and Olson, Franceschetti, and Henkes and Schappert, this pathological picture, which was first described by Leber, is seldom recognized, it is, however, by no means infrequent.

The investigation into the causes of blindness in the Netherlands concerned 227 cases of congenital amaurosis, viz. 114 adult blind patients and 113 children of whom 109 were blind and 4 were partially sighted. Since the Netherlands have approximately 6000 blind persons, 600 of whom are children, these figures mean that the affection is the cause of blindness in 3,8% of all blind persons and in 18% of the blind children.

This affection was erroneously diagnosed by the consulting ophthalmologists as: tapetoretinal degeneration, optic nerve atrophy, (congenital syphilitic) retinochoroiditis and congenital amblyopia (possibly of central origin).

The following symptoms are obligatory:

Congenital blindness or very poor central vision.

In the majority of cases: only very slight fundus changes in comparison with the bad visual function.

Absent or nearly absent electroretinogram.

Recessive hereditary occurrence.

Facultative characteristics are:

Nystagmus—relatively often.

Photophobia—much rarer.

Cataract.

Keratoglobus or keratoconus.

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The two latter symptoms are absent at birth. The affection is described in more detail with the aid of case histories, a characteristic fundus picture and electroretinograms and a first pedigree of 13 cases originating from the Isle of Urk.

Discussion.

Zeeman congratulated Mrs. Schappert on the collecting of such instructive experience regarding this affection, which, on account of apparent infrequency and poverty of symptoms offers great difficulties to the ophthalmologist, but which had been shown to be one of the main causes of blindness in children. The affection deserves our keen attention even after the valuable monograph by Alström. This latter publication seems unsurpassable as a genetic study. As

regards the clinical part, Zeeman remarked—contrarily to Alström's experience “that in no case was there anything observed that could be interpreted as tube vision”—, that he had observed tube vision in two patients.

Tube vision could be even used to advantage, with the aid of so-called telescope glasses, for reading normal print. This fact had remained unnoticed by the speaker in one patient for 15, and in the other for 30 years.

The term “dysgenesis neuroepithelialis retinae” proposed at the time by Waardenburg, may give the clue for the frequent occurrence of keratectasia and cataract among these cases. This term does not only lead to the consideration that the eyecup has not only the potency of later development of a visual function, but also potencies of induction of a lens and of a directional membrane for infiltrating corneal endothelium, as explained by Hagendoorn. Consequently, in a case of dysgenesis neither the development of amblyopia, nor the appearance of lenticular opacities and aberrations of corneal shape are surprising.

Waardenburg: At the time when I read my article in Groningen, I was asked why I considered this abnormality as recessive and rejected an exogenous cause. I had to admit that I had no convincing evidence regarding consanguinity of the parents. My opinion was based on a kind of feeling because of the absence of inflammatory phenomena and the fact, that Leber had fairly often observed consanguinity of the parents, and that I had found consanguinity in two cases. However, a systematic investigation into the matter had not yet been carried out in the Netherlands. I am, therefore, glad that Alström and Olson found consanguinity in convincing multitude in their Swedish material, and that van den Bosch and I have succeeded in demonstrating consanguinity of both parents or between one parent of a blind patient and a parent of another blind patient in a family originating from the Isle of Urk (see pedigree). This example of the rediscovery of Leber's, affection and its considerable frequency also in the Netherlands show how useful Mrs. Schappert's work is. From a scientific point of view, it was an excellent thing that she established contact with the Swedish workers and that Henkes and other colleagues are willing to record the ERG in these patients.

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I understand that Alström wants to exclude all probands with oligophrenia, but this would lead to a haphazard exclusion because, although it is not yet known whether oligophrenia is incorporated in the same gen or has accidentally become manifest together with the amaurosis, there are a considerable number of families in which some affected members are oligophrenic, and others not. For the time being, there is no harm in considering this oligophrenia as a secondary phenomenon, and regarding blindness or marked amblyopia as a criterion. Otherwise, the cases of keratoglobus could also be excluded, because, although I have seen a family in which all the five members of two branches showed amaurosis and keratoglobus, there are several families in which only some members have keratoglobus, and we do not know whether also the other members will develop this affection. The discovery made by Alström and Olson was that patients are not born with keratoglobus.

Van den Bosch, J.: *Ophthalmologica* 137: 422–423 (1959)

A New Syndrome in Three Generations of a Dutch Family.

By 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.