Ophthalmologic and Genetic Study of a Family with Nyctalopia and Myopia

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A large family with nyctalopia and myopia, originating from the province of Zeeland, was subjected to a complete ophthalmological and genetic investigation. The affection proved to be transmitted X-chromosomally and recessively; in none of the obligatory female bearers was any abnormality observed. Of the 23, of course all male patients, 18 were still alive, varying in age between 2 and 70 years. All of them were examined, the youngest two only summarily, however. The clinical picture was highly uniform in all patients. The ocular anomalies shown by them were: (1) subnormal visual acuity of the dominant eye, varying between 0.1 and 0.6, independent of age; (2) myopia varying between 1 and 20 D independent of age and combined with astigmatism in 15 cases; (3) nystagmus was found in 17 patients; (4) strabismus was found in 15 patients, in 9 of them also in a vertical direction, i.e. alternating hypertropia; (5) a dark adaptation study was done in 11 patients; in all cases it was monophasic and there was no measurable scotopic function; (6) apart from the known myopic degenerative disturbances, the fundi showed a characteristic dystrophy of the pigment epithelium in the posterior pole, already entirely distinctly recognizable in the 2- and 4-year-old patients.

The genetic study comprised the determination of possible color vision disturbances and an examination of the blood for the likewise X chromosome-inheriting Xg blood group and G6PD deficiency. Inheritable color vision disturbances were not found, and the hemeralopia with myopia did not lead to measurable abnormalities of color vision. The blood analysis showed that there must be an ample distance between the locus for the Xg blood group and the locus for nyctalopia.

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In addition to the 18 patients, 90 healthy relatives were examined ophthalmologically. No further subjects were found having a myopia with degenerative disturbances, nor persons showing a nyctalopia. The family analysis shows furthermore without any doubt that one gene, or two genes lying particularly closely together on the X chromosome, are responsible for both affections.