A New Pedigree of Night-Blindness Transmitted as an Autosomal Dominant Trait in Chinese

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ABSTRACT

A pedigree of autosomal dominant inheritance of probable congenital stationary night-blindness is described in a Chinese family. The disorder has been transmitted through at least six generations with 21 affected individuals of both sexes.

Congenital stationary night-blindness (CSNB) was one of several dominantly inherited traits in man recognized prior to the publications of Mendel in 1865. The symptoms are characterized by defective twilight vision probablly involving a defect in neural transmission at the level of the rod photoreceptor inner segments and by normal vision in daylight. The most famous pedigree for the night-blindness was that of a Frenchman, Jean Nougaret, who was born about 1637 and who had 135 affected descendents in eleven generations before 1907 (Dell, 1922; Snyder, 1965). Several other pedigrees with fewer members have been reported in Europe and America (Dell, 1922; Carrol and Haig, 1973; Francois *et al.*, 1965; McKusick, 1975). CSNB may also be inherited as an autosomal recessive or as an X-linked form (Carr, 1974). In this paper, a Chinese family with apparent CSNB is described.

CASE REPORT

The propositus (II-2) (Fig. 1) and his ancestors had resided in a small village (Zeu-Chi) and the nearby river basin, Pin-Yang county, in the southeast corner of Che-Kiang province, China, forthree hundred years. He was in general good health, with normal intelligence and normal daylight vision until his death in 1962 at the age of 80. The night-blindness was present and stationary through all of his life without any associated eye anomalies, such as myopia, cataracts or pigmentary changes of the retina. In China, the symptom of night-blindness was also referred to as "chicken eye", which also exhibit lack of vision in twilight. Under ordinary illumination such as candle light or oil burning light the propositus had no visual difficulty, but when he walked in an unlighted place his vision was considerably impaired and did not improve if he remained in the dark. He understood his limitation of vision in darkness and refused to go to outdoor gatherings at night without the aid of a candle lantern or in the company of a person with normal night vision. On may occasions during the 1940's I had accompanied him at night, when I could easily see objects such as steps, ditches, rocks, small animals, etc.; the propositus saw none of these. The night-blindness was one of the major reasons for him and his brother (II-3, also affected) to quit farming and to move out of the village in 1908 to a small town called Sheui-tao, 40 kilometers to

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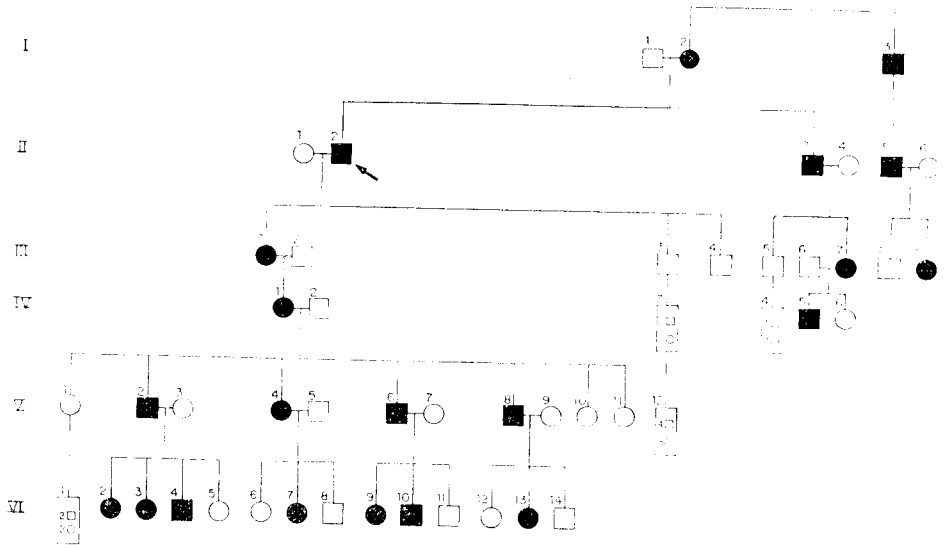

Fig. 1 Pedigree of Chinese family with night-blindness transmitted as an autosomal domina nttrait. Black, affected; open, normal. Arrow indicates the propositus.

the east of the village. During the early part of the century, there was general unrest in the area. Farmers had to partol the fields at night during the harvest time to prevent the loss of grains and livestock thieves. Due to the obvious defect of their visions at night, the brothers could not effectively patrol the fields and protect their property. They finally settled in the town and become seafood retailers, an occupation requiring no night-time work.

It was also learned that the propositus' mother (I-2) and her brother (I-3) had the same symptoms. Six other individuals of the family which I can identify as being affected were two of their daughters (III-1, III-7), the prositus' granddaughter (IV-1), one of her sons (V-2), the propositus' cousin (II-5), and his daughter (III-9). After 27 years of interrupted communication, I have recently contacted one of the propositus' granddaughters (IV-3) who has normal night vision and still lives in the area. She reported to me that several individuals have been born and identified as having night-blindness during the period 1949 to 1975. They are: IV-5, V-4, V-6, V-8, VI-2, VI-3, VI-4, VI-7, VI-9, VI-10, and VI-13 in the pedigree.

DISCUSSION

The mode of inheritance described in this family is autosomal dominant. Men as well as women are affected. There are three examples of male-to-male transmission and all offspring of a normal individual are unaffected with night-blindness. The ratio of normal to affected offspring born to an affected parent was 14:18, which is close to the theoretical value of 1:1.

The symptoms and the mode of inheritance described in this report resemble the historical description of congenital stationary night-blindness originally found in patients from Southern France. A mild form of autosomal dominant retinitis pigmentosa (Deutman, 1977) cannot categorically be excluded by historical evidence alone. Thorough ophthalmological examinations, including dark adaptometry tests and electroretinography, are required to establish a correct diagnosis. Currently, all affected mem-

bers of this pedigree are residing in China, but every effort will be made to examine the patients.

Night-blindnes has always been considered to be an inconvenience and a slight disadvantage to the victims. However, the "inconvenience" may become a severe difficulty and even fatal during wartime or emergency situations. As in other dominant symptoms, etiological basis for the disease is not known.

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中國人的一種體染色體顯性型夜盲症遺傳

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摘　要

本文報告一個在浙南家系的最盲遺傳的調查工作，在這一家系中共有患者21人，男女皆有，而病至少已傳六代之久。由病徵和遺傳方式看，這家系的患者是屬於體染色體顯性型的原發性夜盲症。