

ABNORMAL FUNDUS REFLEXES AND RETINITIS PIGMENTOSA*

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THE normal variation of the fundus reflex which gives a "shot-silk" appearance to the retina, especially in hypermetropes, is well known (Nettleship, 1882; Benson, 1882). Its nature is speculative. A possible significance of grossly exaggerated fundus reflexes was first suggested by Adams Frost (1902) who noticed their presence in two of six female children of a man with retinitis pigmentosa:

The fundus is of the dark type. Everywhere except in the macular region its surface presents a glistening grey white appearance, with a tendency to striation in places. The condition appears to be an exaggeration of the reflex appearance frequently seen round the disc. The vision is normal in all respects.

Niccol (1938) described what appeared to him to be an example of a tapetal reflex in the mother of two children, both of whom had bilateral macular defects. The central regions were stippled by light yellow specks, like gold dust, lying deeper than the retinal blood vessels. While investigating the family of a 30-year-old man with atypical retinitis pigmentosa, Falls and Cotterman (1948) noticed a tapetal-like retinal lustre in the fundi of the patient's mother and in one of his mother's sisters. Scattered on and about the macular area were numerous dust-like golden glittering bodies. On further genetic study, he found other cases both of retinitis pigmentosa and of the tapetal-like reflex. These occurred in such a way as to suggest the operation of a sex-linked gene producing retinitis pigmentosa in males and the tapetal-like reflex in heterozygous females. In this pedigree there was also the suggestion that the chance association or interaction of the genes for the tapetal-like reflex and for colour blindness led to manifest colour blindness in heterozygous females. The responses to the Ishihara test differed from those characteristic of deuteranopia or deuteranomaly. A tapetal reflex has also been described by Mann (1937), but in a woman whose family history is not recorded. More recently, Sorsby (1951) has referred to an appearance resembling a glistening exaggeration of the normal reflex in the fundi of heterozygous females in a pedigree showing sex-linked retinitis pigmentosa. In view of these observations the following case is briefly reported because, although the family investigation was negative with respect to retinitis pigmentosa, the patient has now, 5 years later, herself developed this condition.

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Case Report

A girl aged 9 years first attended the Belgrave Hospital for Children in January, 1949, as her school-mistress suspected that her vision was defective. There was no history of prematurity or of maternal illness during pregnancy.

Visual Acuity: In the right eye 6/36; with +1.5 D cyl. at 140° 6/18. In the left eye 6/36; with +0.25 D sph. +1.75 D cyl. at 35° 6/18. The central areas of the optic fundi, except at the maculae, had a slightly irregular appearance, light being reflected as from the surface of smooth, almost transparent cobble-stones. The optic discs were slightly pale but the vessels appeared normal. There was no abnormality of the periphery of the fundi (Fig.1). The Ishihara test revealed defective colour vision.

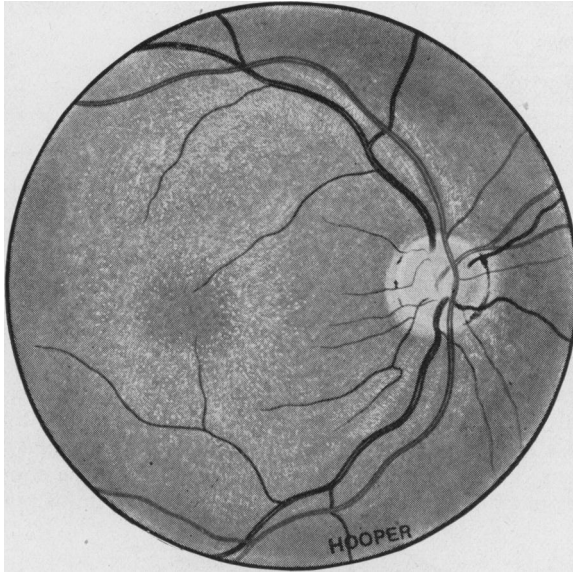


FIG. 1.—Right fundus, 1949.

Professor Arnold Sorsby suggested that such an appearance might indicate the female heterozygous state in sex-linked retinitis pigmentosa and an examination of other members of her family was made. In addition, a detailed investigation of her colour vision was carried out by Mr. W. D. Wright of the Department of Physics, Royal College of Science. He found her very slow with the Ishihara test, she failed on all the charts, suggesting that she had no red-green discrimination, but this was not confirmed with the Farnsworth test in which the discs were arranged roughly in the correct sequence though the detailed ordering showed many errors. There was no one range of the spectrum in which her discrimination was especially good or especially bad. The Nagel anomaloscope and Wright colorimeter tests suggested that she had some colour discrimination although well below normal. She named monochromatic colours correctly (*e.g.*, red, yellow, green, and blue) even when the intensity was altered over a wide range, though the normal protanope or deutanope usually makes a number of mistakes in this. She was not able to make the observations needed to measure her luminosity curve, but her comments about the relative brightness of the different colours did not suggest abnormality in this respect. Mr Wright concluded:

These results have not enabled me to classify her as belonging to one of the recognized groups of colour defect. I can only say that her colour discrimination is generally poor

but not non-existent, and I would guess that her defect was associated rather with her discrimination capacity than with any abnormality of her colour-perception mechanism. Whether this means that her defect is located in the retina or in the higher centres rather depends on one's ideas of where discrimination occurs. I would not care to speculate on this.

An examination of 28 of the 38 living members of the child's family was carried out with completely negative results (Fig. 2). No abnormal fundus appearances or defective colour vision were discovered. All had good corrected visual acuity in each eye except for an aunt with one amblyopic eye from neglected convergent squint. The relatives of the normal father were examined as part of a systematic attempt to discover any possible genetic significance for the condition.

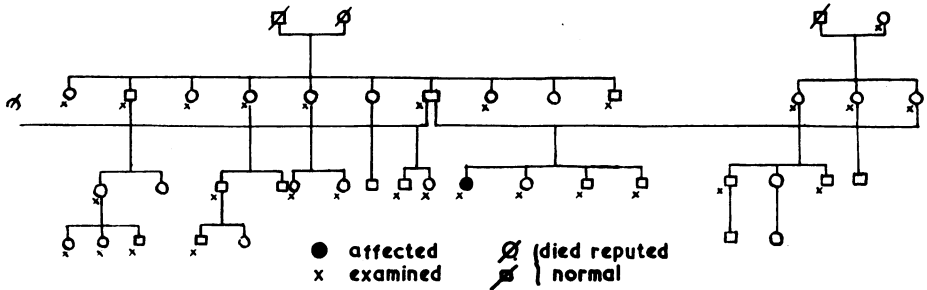


FIG. 2.—Family pedigree.

On routine examination 3 years later the findings were unchanged; 5 years after her first attendance she was seen again because her brother had noticed that when playing with a ball, she sometimes did not know where it had gone. There was no complaint of difficulty in twilight vision. Her corrected visual acuity was still 6/18 in each eye, but her optic discs had a translucent pallor, the retinal vessels were slightly attenuated, and pigment aggregations of the bone corpuscle type were seen in the equatorial region of each fundus. The ophthalmoscopic appearance was that of retinitis pigmentosa (Fig. 3).

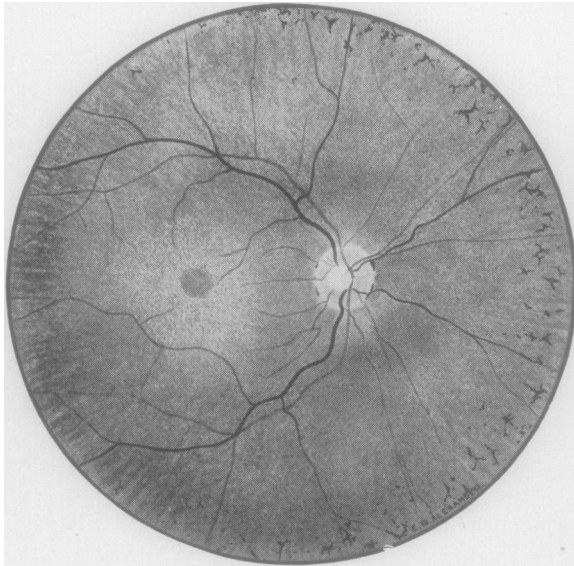


FIG. 3.—Right fundus, 1954.

The records of her fields of vision were somewhat unsatisfactory as she found difficulty in concentrating to maintain fixation. The peripheral fields showed only limitation in their upper parts more marked on the temporal side, but the inner isopters had a general restriction on the temporal side of the field of vision of each eye (Fig. 4).

L.V. 6/36
 OBJECTS 6/330, 7/2000 WHITE
 18-8-54
 R. P. C.

R.V. 6/36
 OBJECTS 6/330, 7/2000 WHITE
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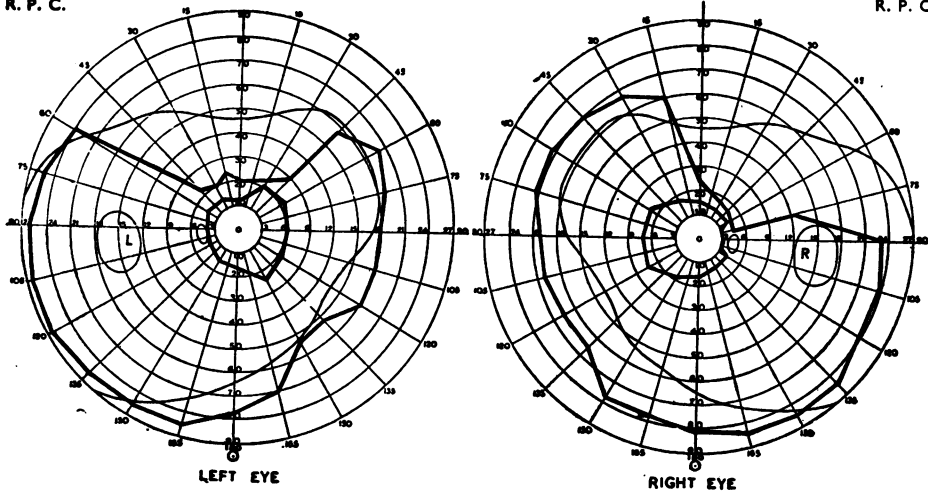


FIG. 4.—Visual fields, 1954.

Dark-adaptation tests revealed impairment of both the cone and rod parts of the curve (Fig. 5).

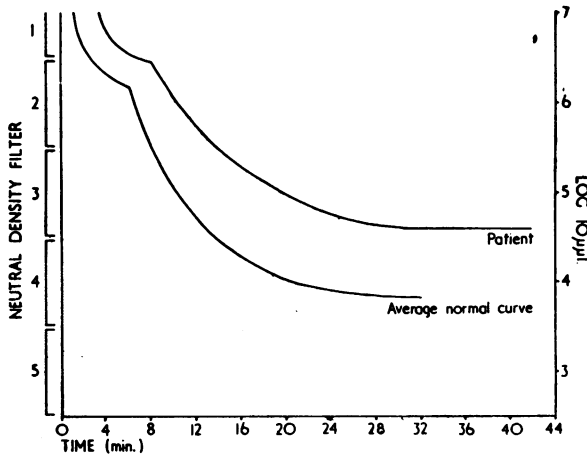


FIG. 5.—Dark-adaptation curve, 1954, compared with average normal curve recorded under same conditions.

The ocular media were clear, an x ray of the skull was reported normal, her hearing was good, and there were no other abnormalities.

Discussion

With the completely negative family history dominant inheritance is clearly excluded, similarly recessive sex linkage is ruled out if only because of the sex of the patient. There is nothing in the pedigree (Fig. 2) to support the possibility of autosomal recessive inheritance for the parents were not consanguineous. One can only speculate that a mutation for retinitis pigmentosa has occurred in the x-chromosome of the germ cells of one of the parents and that the patient is potentially the mother of a family in which sex-linked intermediate retinitis pigmentosa may occur. It is known that some heterozygous females may show the characteristic carrier condition such as a tapetal reflex while others may occasionally show the disease itself (Roberts and Sorsby, personal communication). Full expression of a recessive pathogenic gene in the carrier female is not particularly uncommon in several x-chromosomal affections such as colour anomalies and sex-linked nystagmus.

Summary

A case is reported in which a grossly exaggerated fundus reflex was associated with impaired visual acuity and a lack of colour discrimination which did not belong to one of the recognized groups of colour defect. Despite evidence suggesting that the condition might be an indication of the female heterozygous state in intermediate sex-linked retinitis pigmentosa, no ocular abnormality was found in 28 of 38 living relatives and the pedigree did not support any of the usual types of inheritance. Five years later, however, the patient presented with clear signs of retinitis pigmentosa. It is concluded that a mutation has occurred in the x-chromosome of the germ cell of one of the parents, and that the patient is in fact probably heterozygous for retinitis pigmentosa showing clinically at first a tapetal reflex and later the picture of the disease itself.

A knowledge of the manifestations of the heterozygous state in genetic disease is of clinical importance both for diagnosis and prognosis. Further work on this ill-defined group of fundus reflexes may endow them with a more exact significance.

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