Editorial: Retinitis pigmentosa

Retinitis pigmentosa is a term commonly used to describe a genetically determined group of disorders characterized primarily by a progressive deterioration in the function of the rods. Most cases ultimately result in blindness and the group as a whole is responsible for over 6% per cent. of all the registered blind in England and Wales up to the age of 65 years (Sorsby, 1972).

The hereditary nature of retinitis pigmentosa has been recognized for over one hundred years and, particularly since the pioneer studies of Nettleship (1907-8) and Julia Bell (1922), an enormous amount of work has been published about the patterns of inheritance of this group of disorders. It has long been recognized that retinitis pigmentosa can be transmitted as an autosomal dominant, an autosomal recessive, or an X-linked trait, the last often being subdivided into recessive, intermediate, and dominant X-linked forms. Almost without exception, all large series of cases of retinitis pigmentosa show a male preponderance, the incidence in males being variously quoted as between 55 and 63% per cent. of all cases. This excess of males has generally been explained by the occurrence of the X-linked form of the disease, yet until recently X-linked retinitis pigmentosa was considered to be by far the rarest genetic form of the disease (François, 1961—4·5 per cent.; Ammann, Klein, and Franceschetti, 1965—1 per cent.). The recent study at Moorfields Eye Hospital, London, culminating in the report by Bird in this issue (p. 177) has demonstrated that the X-linked form of the disease accounts for over 20 per cent. of all cases of retinitis pigmentosa in England, a figure which is consistent with a male incidence of about 60% per cent. Bird has also shown that, in X-linked retinitis pigmentosa, all adult heterozygous females demonstrate some retinal abnormality. In some cases these changes are florid, in others subtle, but as a result of these findings there now appears to be little justification for subdividing X-linked retinitis pigmentosa into recessive, intermediate, and dominant forms.

There are practical reasons for recognizing the different genetic forms of retinitis pigmentosa. Firstly, it has been shown that the different genetic forms of the disease have different visual prognoses (Jay, 1972). The majority of patients with autosomal dominant retinitis pigmentosa will retain a central vision of 6/18 or more at 50 years of age. In autosomal recessive retinitis pigmentosa, and in affected males with the X-linked form of the disease, the majority of patients have a central vision of 6/60 or less at 30 years of age. Secondly, in order to give accurate genetic counselling, it is necessary to determine the mode of inheritance in the family in question. By his accurate description of affected males and heterozygous females with X-linked retinitis pigmentosa, Bird has made possible the recognition of isolated cases of this condition, particularly when an affected male can be shown to have a mother or daughter with the features of the heterozygous state. The recognition of heterozygous females also permits the selective abortion in early pregnancy of their male fetuses, half of whom can be expected to suffer from a severe form of retinitis pigmentosa.

There are also theoretical reasons for recognizing the different genetic forms of retinitis pigmentosa. Although so far little progress has been made in our understanding of the underlying defects in this group of disorders, it is only by their accurate delineation that any progress can be expected to occur, for each genetic form can be expected to result from a different basic abnormality. It would not be surprising if retinitis pigmentosa followed the general pattern for genetically determined disorders (a pattern which does, however, have exceptions), wherein dominantly inherited traits tend to be structural anomalies, whereas recessively inherited traits tend to be inborn errors of metabolism, for example, enzyme defects (McKusick, 1972). It has been suggested that, in autosomal dominant retinitis pigmentosa, there may be a reduction in the amount or effectiveness of rhodopsin in each rod throughout the retina (Berson, Gouras, and Gunkel, 1968), and
that the receptors in retinitis pigmentosa may be reduced in length (Highman and Weale, 1973). These findings could be interpreted as indicating a structural anomaly in the rods, an interpretation that does, however, require histological confirmation in the early stages of the disease. In autosomal recessive and in X-linked retinitis pigmentosa, the possible demonstration of an enzymatic defect still lies in the future, but it would not be unreasonable to expect such a defect to be situated in the pigment epithelium of the retina. Females heterozygous for X-linked retinitis pigmentosa, whose disorder is usually particularly mild, may be valuable models for further study into the aetiology of this condition.

References

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