Editorial: Small optic discs

The responsibility for the detection of a small optic disc lies solely within the sphere of the ophthalmologist. Early recognition provides a service to the paediatrician and probably also to the neurologist, as the following papers in this issue stress. Most importantly, however, it helps the patient by obviating the need for further investigation and directing future management towards the endocrinologist and other specialists.

Optic nerve hypoplasia was thought by Cords (1923) and other earlier writers to be extremely rare. Indeed, Scheie and Adler (1941) were able to cite only 5 cases apart from their own. It has recently been recognised with increasing frequency in 20 or more cases (Walton and Robb, 1970; François and de Rouck, 1976).

The diagnosis of hypoplasia presents little difficulty in the extreme case. The disc is small, with slightly tortuous and occasionally small vessels and is surrounded by a yellowish mottled peripapillary halo with a pigmented rim approximately corresponding to the size of a normal disc (Ridley, 1938; Scheie and Adler, 1941; Whinery and Blodi, 1963; Walton and Robb, 1970). On both ophthalmoscopic and pathological examination the retinal nerve fibre layer is seen to be thinned (Whinery and Blodi, 1968; Manor and Korczyn, 1976). The condition is often bilateral, may be asymmetrical, and central vision is usually involved. However, field defects are variable and include bitemporal hemianopia (Ellenberger and Runyan, 1970), unilateral temporal hemianopia (Seeley and Smith, 1972), and some patients may have abnormalities of ocular movement, including see-saw nystagmus (Davis and Schock, 1975).

Frisén and Holmegaard in this issue present an interesting account of 7 patients with degrees of hypoplasia ranging from the classical, functionally devastating type to ones with a defect so mild as to go unnoticed by all but the most experienced developmental neuro-ophthalmologist. Frisén even believes that hypoplasia may occur in discs of normal size when one takes into account the variability of normal disc size and diffuse loss of the retinal nerve fibres; in his opinion hypoplasia therefore ranges from severe (where the disc diameter fails to reach the normal lower limit) to undetectable (with present tools). Not all would agree with the extent of this concept, however, and it would appear that there is a distinction based on embryological grounds, the smaller discs occurring as a result of an injury early in embryogenesis, the ones with normal sized discs being the result of a later even postnatal injury.

The important association between hypoplastic discs and cerebral malformation was first clearly suggested by de Morsier (1956), although Reeves (1941) had previously described a case with congenital absence of the septum pellucidum associated with "bilateral primary optic atrophy of undetermined origin, probably, however, on the basis of congenital aplasia". De Morsier (1956) named this condition septo-optic dysplasia, and later Hoyt et al. (1970) described 8 patients with optic hypoplasia and congenital hypopituitarism, 3 of whom showed absence of the septum pellucidum. In 1972, 2 groups from the Hospital for Sick Children, Great Ormond Street, namely, Brook et al. (1972), and Harris and Haas (1972), pointed out that the onset of slowing of growth may be delayed until 18 months to 3½ years. Billson and Hopkins (1972) reported 2 similar cases, one of which had the endocrinological and ophthalmological features of septo-optic dysplasia but had a normal septum pellucidum. This finding was confirmed by Patel et al. (1975).

Other cerebral and facial malformations, mainly of mid-line structures have been reported (Patel et al., 1972; de Myer et al., 1964; Hale and Rice, 1974), and there may be a relationship with the Kallmann syndrome (Kallmann et al., 1943), in which hypopituitarism and anosmia may be associated with forebrain abnormalities.

In this issue Hoyt and Billson describe an association between the maternal ingestion of anti-convulsants with known teratogenic properties (phenytoin) and optic nerve hypoplasia in the child. Patel et al. (1975) found that of the 4 patients they described 2 had a prediabetic mother, and Petersen and Walton (1977) described 17 children, born of severely diabetic mothers, who had what was described as segmental optic nerve hypoplasia; this term is discussed by Dorrell in this issue. Maternal ingestion of quinine has also been indicated as a cause of optic nerve hypoplasia (McKinna, 1966), and in cattle viral infection has been implicated (Bistner et al., 1973).

The relationship of hypoplastic discs to 'tilted' and segmental optic nerve hypoplasia (Petersen and Walton, 1977) is not clear, but some useful specu-
lations are made by Dorrell in later pages of this journal.

A number of authors believe that total aplasia of the optic nerve occurs as a result of failure of incorporation of mesoderm into the optic stalk, which normally occurs at the 4 to 10 mm stage. Hypoplasia, however, results from a defect in the normal differentiation of the retinal ganglion cells at the 13 to 15 mm stage or a failure of their processes to reach the optic stalk by the 19 mm stage. The anterior wall of the diencephalon forms the lamina reuniens between the 16 and 22 mm stage. It is from this area that the olfactory bulbs and later the corpus callosum and septum pellucidum, among other structures, arise. Septo-optic dysplasia may reflect a dysfunction of the anterior wall of the diencephalon or an independent injury to the optic vesicle and the anterior diencephalon.

This issue provides a series of thought-provoking papers on subjects which have previously largely escaped recognition. The nosological, aetiological, and clinical implications are of importance to us all.

References


