Lenticonus in spina bifida

A case report

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Anterior lenticonus is a rare anomaly, first described by Webster (1874), which is frequently associated with Alport's syndrome of nephritis and perceptual deafness. A report of the 42 cases recorded up to 1966 is given by Arnott, Crawford, and Toghiani, 1966.

South Wales has an unusually high incidence of spina bifida. The incidence was found to be 3.92 per 1000 in Glamorgan and Monmouthshire with approximately 30 000 births per year (Richards and Lowe, 1971). The reason for this is unknown and it was of interest to find a case of spina bifida having anterior lenticonus with other multiple congenital abnormalities.

Case report

The female infant weighed 1078 g and lived for 3 days, being the first child of a 21-year-old woman. There was no history of infection or drug therapy during pregnancy.

CLINICAL FINDINGS

The head was grossly abnormal in shape with low set ears. Hypertelorism and mongoloid palpebral fissures were present. There was a right-sided hare lip and cleft palate. The spina bifida was extensive with a flat lumbar myelocoele (T10-S2 segments).

NECROPSY FINDINGS

The heart was moderately enlarged and the ductus arteriosus still patent despite the absence of other abnormalities. The uterus was slightly bifid. There was a partial deficiency of the cranial bones at the vertex. The cerebral hemispheres were small with interdigitations through deficiencies in the falx. The corpus callosum was absent with a radial pattern of gyri from the hilus of the hemispheres. There was an Arnold-Chiari malformation and a compressed ventricular system with obliteration of the basal cisternae. All cranial nerves were identified.

Study undertaken in the Department of Ophthalmology, during the author's final year as an undergraduate

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OPHTHALMIC FINDINGS

The eyes were microphthalmic. There was bilateral asymmetrical anterior lenticonus with posterior concavity (Figs 1 and 2). The iris and ciliary body were fully developed in both eyes. The optic nerves were very small and there was aplasia of the optic nerve head with no vessels emanating from it to the retina.

Chromosome studies were attempted but cell cultures failed.

Discussion

Anterior lenticonus has not been described previously as part of a multiple congenital abnormality syndrome. The majority of reported cases of anterior lenticonus have been bilateral and present in males. Alport's syndrome is present in approximately 20 per cent of cases (Arnott and others, 1966). Anterior lenticonus appears to be an inherited characteristic of this syndrome the mode of inheritance of which is autosomal dominant.

There have been no females with anterior lenticonus reported in Alport's syndrome and only three females in all 42 cases reviewed (Arnott and others, 1966). A sex-linked modifying gene has been suggested to explain the different sex incidence in Alport's syndrome and this would appear to be influencing anterior lenticonus formation in all cases.

Although most cases of isolated anterior lenticonus have been sporadic, further evidence of a genetic causation has been given by Urrets Zavala and Obregón Oliva (1939) who described two cases who were brothers and whose parents were cousins.

Stevens (1970) reported a case of anterior lenticonus in Waardenburg's syndrome, which has an autosomal dominant mode of inheritance. Similarities between Waardenburg's and Alport's syndromes are normally perceptual deafness and an autosomal dominant mode of inheritance, but in two cases arcus juvenilis and anterior lenticonus also occurred.

The multiple congenital abnormalities present with this case of lenticonus are of considerable

FIG. 1 (left) Lens showing anterior lenticonus

FIG. 2 (right) Lens showing unusual posterior dimple

interest. The cause of spina bifida is uncertain. Although it is widely thought to be multifactorially inherited, the triggering mechanism for its expression is unknown. All cases of spina bifida may be classified either as isolated or as part of a multiple congenital abnormality syndrome (which may involve the eyes). This association with anterior lenticonus suggests, on present knowledge, a clear-cut preuterine causation. This is further substantiated by the association with low set ears and cleft lip and palate which are often present with chromosomal defects.

Altogether 356 infants with spina bifida were born between 1964 and 1966 in Glamorgan and Monmouthshire (Richards and Lowe, 1971). Multiple congenital abnormalities were associated with spina bifida in 31 of these (personal observation). Only two had macroscopic eye defects consisting of anophthalmos in one and hypotelorism in another. Previously described eye defects include: anophthalmos, microphthalmos, colobomata, absence of the optic nerves and ganglion cells of the retina,

and absence of the macula (Walsh and Hoyt, 1969). Anophthalmos and colobomata are known to occur as genetic abnormalities (Waardenburg, Franceschetti, and Klein, 1961).

This particular case of anterior lenticonus is unusual as it had a posterior dimple and a conus involving the whole anterior surface of the lens. The presence of a posterior dimple is an extremely rare condition. Only one other case has been reported (Mohr, 1910). This was a child, 3 weeks old, with an enlarged eye and raised intraocular pressure.

Summary

A previously unreported association between spina bifida and anterior lenticonus is noted. The significance of this finding to the aetiology of some cases of spina bifida is discussed.

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