Atavistic ocular ossicle

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Metaplastic ossification frequently follows ocular scarring. Anterior scleral ossicles are normal in most common fish, *e.g.* dogfish, and in reptiles, and are said to have accommodative and protective functions. Birds (219 out of 532 species: Tiemeier, 1950) have an os opticus of highly cancellous texture lying in the posterior sclera and surrounding the optic nerve head in one or several places. Duke-Elder (1958) wrote that "no satisfactory theory for its presence has been put forward". He also mentioned the occurrence of "fatty tissue, cartilage, and even bone" in coloboma of the vitreous in microphthalmic eyes (Duke-Elder, 1964). This description may refer to cases now being recognized as examples of the 13-15 trisomy syndrome. Saraux and Dhermy (1968) found a nodule of well-developed bone in the line of a uveal coloboma but located episclerally near the optic nerve. This was present in a case of D-trisomy with translocation.

This paper reports a case of congenital isolated posterior intrascleral ossification, which appears to be a unique finding.

Clinical history

A married woman aged 23 years was delivered at full term of her first baby on May 25, 1969. She had had "tablets" for hyperemesis gravidarum in the first trimester and apart from an *E. coli* pyelitis, the antenatal history was uneventful. She was blood group O RH+. The Wassermann reaction and Venereal Disease Reference Laboratory tests were negative. There was no family history of ocular disorder. The baby girl, delivered by vacuum extraction and weighing 3.3 kg., was admitted to the Special Care Unit where she was found to have a left facial palsy and bilateral colobomata. She died 48 hours later after repeated convulsions. Skull and lateral spinal x rays, urinary chromatography, and karyotyping gave normal results. Laboratory tests showed evidence of urinary infection and the serum had low calcium, raised phosphorus, and normal electrolyte levels.

Pathology

AUTOPSY FINDINGS

The baby weighed 3 kg. The cranium and face appeared to be normally formed except for atypical dysplasia of the left auricle, which had a large flat helix with a single layer pointing upwards and an absent lobe. The right ear was large but had a tiny lobe. The eyes appeared to lie within normal orbital fossae. No congenital defects were found in the other systems. The cause of death was bilateral bronchopneumonia.

Macroscopical examination

The left eye measures $20 \times 19 \times 18$ mm. with an indefinite cornea of approximately 10 mm. diameter and an inferior nasal iris coloboma. The opened specimen shows a

typical but irregularly-shaped coloboma involving the retina and uvea and extending just to include the disc.

The right eye measures $19 \times 18 \times 17$ mm. The corneal diameter is 9 mm. and the anterior aspect shows an inferior nasal coloboma. The opened specimen shows an irregular coloboma of the uvea and retina extending above the disc, which is itself abnormal. The total area involved by maldevelopment is greater than in the left eye.

The external ocular muscles appear to be normally developed on both sides.

Microscopical examination

The left eye shows no abnormality of the cornea. The coloboma involves the iris, ciliary body, retina, and choroid in varying degrees, with the conventional histology seen in this defect. The iris tissue near the coloboma shows persistence of a primitive type of mesoderm. The lens appears to be normal. There is slight ectasia of the sclera along the line of the embryonic fissure, and about 3 mm. from the optic disc there is a focus of cartilage proceeding to ossification (Figs 1 and 2) lying encased by scleral mesoderm at all levels. The normal constituents of bone are present with osteoid tissue, osteoblasts, an occasional osteoclast, Haversian canal systems, and scattered normoblasts (Fig. 3). The globe has been serially sectioned and the ossicle measures 0.3 mm. in thickness. No choroidal elements have formed in this area and the inner scleral layers covering the bone are in direct contact with attenuated retina which has an occasional rosette-like formation. Myelination of the optic nerve is normal for the age of the infant.

FIG. 1 *Intrascleral chondro-ossification in uveal coloboma with overlying attenuated dysplastic retina. Haematoxylin and eosin. $\times 8$*

FIG. 2 *Area of dense compact bone and periosteal-type cells. Haematoxylin and eosin. $\times 330$*

FIG. 3 *Ossifying zone with an osteoclast and marrow formation. Haematoxylin and eosin. $\times 85$*

The right eye shows scleralization of the cornea which is otherwise normal. The coloboma involves the iris, ciliary body, choroid, and retina to varying degrees. The choroid has failed to develop in the posterior part of the embryonic fissure and this defect extends across the optic disc to the macula. The lens is not remarkable. The iris shows irregular hyperplasia of the pigment layer, particularly at its root. Dysplastic retinal tissue lies in direct contact with ectatic sclera and shows focal herniation of retinal elements into orbital fat. A coloboma of the optic disc is associated with excavation of the sclera, the lumen of the cavity being filled with whorls and fasciculated optic nerve fibres. These fibres are degenerate and show corpora amylacea. No cartilaginous or bony elements have been found on serial sectioning. The optic nerve is not remarkable.

Comment

Colobomata of the eyes, microphthalmos, and anomalies of the ears have been described in association with paralysis of the VIIth nerve. This is said to be due to central atrophy of the nucleus (nuclear aplasia: Möbius, 1892). It is possible that the present case is a variant of Möbius's syndrome.

Intracartilaginous ossification occurred in the sclera of this eye. Cartilage is normally present in the posterior sclera of certain fish and reptiles. It is found in the severely microphthalmic colobomatous eyes of the 13-15 trisomy syndrome, its location being in the anterior para- or retrolental area near the coloboma. Manschot (1958) mentioned the presence of retrolental cartilage in a case of persistent hyperplastic vitreous. Yanoff and Font (1969) found cartilage in the vitreous in unilateral microphthalmos in a normal 4-year-old girl, and Garner and Griffiths (1969) reported bilateral cartilaginous plaques in anterior staphylomatous tissue in a foal. Published papers to date have shown cartilage in the anterior half of the globe in the human eye. Dhermy (1970), however, found an intrascleral island of cartilage between a scleral coloboma and the optic nerve in a case of a ring-13 chromosome.

As far as can be ascertained no case similar to the one now presented has been described. The cause of the scleral ossification in the left eye could be sequestration of a focus in the embryonic fissure from the neighbouring orbital chondrocranium. It appears to be more likely, however, that the primitive mesenchyme forming the sclera had undergone heteroplastic changes, *i.e.* a primary anomalous differentiation in the developing tissue. The paraxial mesoderm which forms the scleral coat extends from the limbus to reach the posterior pole at the 5th month of intrauterine life. Thus the ossification can be presumed to have occurred during the last 4 foetal months. The proximal end of the embryonic fissure failed to close in the right eye but no bone or cartilage was found.

Having regard to the points of similarity between the scleral bone in the present case and the os opticus of lower species, it is tempting to call it an atavistic ossicle.

Summary

Isolated posterior intrascleral ossification in a colobomatous eye is described in a full-term infant with normal karyotype. Death occurred 48 hours after delivery. The formation of such an ossicle is thought to be a unique occurrence.

We wish to acknowledge the help of Dr. E. Dillon, who performed the autopsy, Dr. J. Masterson for karyotyping, and Mr. R. Lester, F.I.M.L.T., for technical aid.

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