CASE REPORTS

A case of amniotic band syndrome with bilateral epibulbar choristoma

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Abstract

An autopsy case of amniotic band syndrome with bilateral epibulbar choristoma is described. The left eye reveals a complex choristoma and the right eye a dermis-like choristoma. Both choristomatous lesions included lenticular tissue suggesting that rupture of the amnion, which is the initial event of amniotic band syndrome, might have occurred at about the fourth week of gestation. Since the other systemic manifestations of amniotic band syndrome are considered to be compression deformities of the fetus caused by oligohydramnios or amniotic band, the occurrence of epibulbar choristomas in both eyes in this case suggests that a compression mechanism may play a role in the pathogenesis of epibulbar choristoma.

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Early rupture of the amnion, leading to oligohydramnios and formation of the amniotic constrictive band, is considered to be the initial event of amniotic band syndrome, resulting in compression deformities of the fetus. The manifestation of intrauterine compression, such as amputation of the extremities and digits, cranio-facial clefts that often do not correspond in location to disturbances in normal embryogenesis, are diagnostic of amniotic band syndrome.¹ Ocular tissue may also be involved. Common ocular manifestations are congenital or acquired corneal opacity, ectropion, coloboma of eyelid, microphthalmos, strabismus, and hypertelorism.²⁻⁴ Ben Ezra *et al*⁵ reported a case in which a uveal coloboma was probably caused by an amniotic band that had lodged in the fetal fissure and prevented the closure. We describe an autopsy case of amniotic band syndrome which presented with corneal opacity, ectropion, and coloboma of the eyelid, as well as other systemic deformities. We report the histological features of the globes and eyelids of this uncommon malformation.

Case report

A 28-year-old Japanese woman gave birth to a 565 g female neonate at 25 weeks of gestation by spontaneous vaginal delivery. The woman had aborted twice and this pregnancy was complicated by an ultrasonographically suggested acrania probably due to amniotic band syn-

Figure 1 The scalp of the fetus and amnion merge at the supraorbital region and the head is attached to the placenta. P=placenta; U=umbilical cord; V=viscera.

drome. Ultrasonography demonstrated oligohydramnios and adherence of the head of the fetus and the placenta. The level of α fetoprotein was elevated both in the maternal serum (2034 μ g/l) and the amniotic fluid (238 480 μ g/l; normal level >20 μ g/l). The Apgar score was 0 at 1 minute and the neonate could not be

Figure 2 The face of the fetus. Placenta and brain were removed. Left eye reveals ectropion and coloboma of the lower lid. The right eye shows that the palpebral fissure is scarcely open.

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resuscitated because of extensive craniofacial deformities.

A fibrous band was attached to the scalp of the fetus and inspection revealed multiple malformations. The skull bones were incompletely formed and brain tissue was exposed. The scalp was contiguous with the amnion so the head of the fetus was attached to the placenta. Other systemic abnormalities were as follows: abdominal and right thoracic wall defects, coarctation of the aorta, amputation of the third and fourth fingers of the left hand, amputation of the first, second, fourth, and fifth toes of the left foot, facial cleft, scoliosis, and short umbilical cord (Fig 1).

Other malformations observed by inspection were as follows: right eye, the palpebral fissure was so narrow that the right globe was obscured; left eye, ectropion and coloboma of the lower

eyelid were present. An opaque cornea-like structure was observed and was contiguous with upper eyelid, causing incomplete formation of the upper conjunctival fornix (Fig 2).

Histopathology

RIGHT EYE (FIG 3A)

Both the upper and lower eyelids were well formed showing eyelashes, the extraocular muscles, and tarsus composed of collagenous fibres including Meibomian glands. The cornea, anterior chamber, and iris were replaced by dermis-like connective tissue including rudimentary lens, cartilage, hair follicles, and pilosebaceous units. This lesion was covered with stratified squamous or cuboidal epithelium (Fig 3B). The posterior segment was almost normal except for the dysplastic retina showing rosette formation. The pigment epithelium did not reach the optic disc resulting in the dysplastic retina lying in direct contact with the sclera, suggesting an incomplete closure of the fetal fissure. The hyaloid artery was observed (Fig 3C).

LEFT EYE (FIG 4)

The left eye showed similar histological findings to those of the right eye except for minor

Fig 3A

Fig3B

Fig 3C

Figure 3 (A) Right globe and eyelids. The anterior segment is replaced by fibrous tissue containing cartilage (arrowhead), lenticular tissue (asterisk), and skin appendages (arrow) (haematoxylin and eosin, $\times 2.5$). (B) The anterior segment of right eye. The fibrous tissue includes skin appendages (haematoxylin and eosin, $\times 150$). (C) The retina with dysplastic changes showing rosette formation lying directly on the sclera. Retinal pigment epithelium fails to reach optic disc (arrow) (haematoxylin and eosin, $\times 100$).

Fig 4A

Fig 4B

Figure 4 (A) The anterior segment of the left eye is also replaced by fibrous tissue (asterisk) (haematoxylin and eosin, $\times 2.5$). (B) The fibrous tissue includes lenticular tissue (arrow), but no skin appendages (haematoxylin and eosin, $\times 100$).

differences. The lower eyelid was well formed but the upper eyelid was rudimentary. The cornea, anterior chamber, iris, and ciliary body were replaced by fibrous connective tissue (Fig 4A). This connective tissue was covered with stratified squamous epithelium and included islands of lenticular tissue, but no other components such as skin appendage and cartilage were found (Fig 4B). The posterior segment was generally unremarkable.

Discussion

The incidence of amniotic band syndrome has been estimated at one in 5000 to 15000 live births. Deformities observed in amniotic band syndrome have been considered to be compression deformities caused by the amniotic band or amnion itself⁶; the orbit or globe may be involved.

Epibulbar choristomas can be divided into four main histopathological groups; dermoid, lipodermoid, single tissue choristoma, and complex choristoma.⁷ A dermoid consists of dermis-like connective tissue in which pilosebaceous units are present. When the lesion consists of dermis-like connective tissue and no pilosebaceous units it is termed a dermis-like choristoma, belonging to single tissue choristoma.⁸ Complex choristomas consist of two or more choristomatous tissues.⁷ In the left eye of the present case, the cornea, anterior chamber, and iris were replaced by choristomatous dermis-like connective tissue covered by stratified squamous epithelium with hair follicles, pilosebaceous units, and a cartilaginous tissue. From these findings the left eye was diagnosed as an epibulbar complex choristoma. The right eye of the present case showed similar histological findings to those of left eye. The cornea, anterior chamber, and iris were replaced by choristomatous dermis-like connective tissue covered with stratified squamous epithelium. Since no other choristomatous component could be found the right eye was diagnosed as dermis-like choristoma.

In both eyes alterations in the posterior segment were milder compared with the anterior segment. In the left eye focal dysplastic changes

of the retina showing rosette formation were seen. We have previously reported two cases of corneal choristoma and both of them showed similar retinal dysplastic change.⁸ In both eyes lenticular tissue is intermingled in the choristomatous tissue, suggesting that the malformation of the eye occurred during formation of the lens at about the fourth week of gestation,⁹ and that the rupture of the amnion in this case happened at about the same time or before.

There is no satisfactory explanation of the origin of the epibulbar choristoma.¹⁰ Ozanics¹¹ suggested that many congenital malformations of the anterior segment of the eye probably arose from derangement in the migration of neural crest cells. The case described here could present evidence to support this theory because the choristomatous tissue consists of cartilaginous and fibrous tissues which are derivative of neural crest cells. So the fact that this case of amniotic band syndrome had epibulbar choristoma in both eyes might indicate that a compression mechanism could play a role in the pathogenesis of epibulbar choristoma by disturbing the migration of the neural crest cells into the optic cup.

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