ENCANTHOSCHISIS*

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THE caruncle receives little attention in ophthalmic literature and abnormalities are rarely reported. A supernumerary caruncle has been reported by Eyre (1897), Stephenson (1896), and Schreiber (1913). A split caruncle is very rare and occurs with a group of abnormalities at the inner canthus, to which the term encanthoschisis was given by Hirsch (1912).

The following case of encanthoschisis shows the incomplete form of mandibulo-facial dysostosis associated with the Klippel-Feil syndrome.

Case Report

A child aged 7 years shows a slight anti-mongoloid slope of the left palpebral aperture (Fig. 1).

FIG. 1.—Child's appearance.

The outer canthus of the left eye (Fig. 2, opposite) shows a defect in which lashes and tarsal plate are absent. The aperture of the left inner canthus is horse-shoe shaped, in contrast to the normal acute angle seen on the right side. The left caruncle is divided into two almost equal parts and a thin band of glistening fibrous tissue passes between these two halves of the caruncle and extends from the skin to the conjunctiva (Fig. 3, opposite).

There is a wedge-shaped defect of the plica semilunaris. There is a marked alternating convergent strabismus. The right palpebral aperture, caruncle, and plica semilunaris are normal.

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FIG. 2.—Defect in outer canthus of left eye.

FIG. 3.—Fibrous tissue in left caruncle.

General Examination.—There is a mild degree of maxillary and mandibular hypoplasia on the left side and an accessory auricle is present on the left side. There is also a marked skeletal deformity, associated with fused cervical vertebrae, webbed neck on the right, and marked scoliosis.

Discussion

As already mentioned, encanthoschisis is the term used by Hirsch (1912) to describe certain abnormalities at the inner canthus, which are also present in this case, and whose main feature is a split caruncle. The full syndrome comprises:

1. A caruncle split into two more or less equal parts and separated by a band of fibrous tissue.
2. A horse-shoe shape to the aperture of the inner canthus instead of the normal acute angle.
3. A defect of the plica semilunaris.

Hirsch (1912) described the first and, I believe, the only previous case of this syndrome, a 14-year-old girl, in whom the syndrome was bilateral. She also had other congenital defects, notably accessory auricles, a defect in her jaws and incisors, wedge-shaped prolongation of the hairline, and micrognathia; it is clear that this patient also had mandibulo-facial dysostosis.

Hirsch described another case, a 40-year-old mason, in whom he thought incomplete encanthoschisis was present. This patient did not, however, have mandibulo-facial dysostosis.

It is interesting that the present case also has mandibulo-facial dysostosis, and it seems probable that encanthoschisis is more likely to occur with this syndrome. Although more than 150 cases of mandibulo-facial dysostosis have been reported, abnormalities of the caruncle have not been described and are not mentioned by Franceschetti and Klein (1949), François and

Haustrate (1953), Thier (1959), and Franceschetti, Klein, and Brocher (1959). However, in some of the illustrations of published cases, a horse-shoe shaped inner canthus can be seen and it is possible that encanthoschisis may be present in some of these cases.

As previously stated, a split caruncle, such as occurs in encanthoschisis, is likely to occur in association with mandibulo-facial dysostosis, which most authors think to be genetically determined. Mann and Kilner (1943) stated that this was caused by a malformation of the maxillary mesoderm in the embryo, and it is now generally agreed that the syndrome is caused by arrested development of the first branchial cleft at about the seventh week of gestation.

Franceschetti and Klein (1949) believed that it was caused by a disturbance of the action of the organization centre due to a lethal or sublethal effect on the gene concerned. Weyers (1951) suggested that infections of the mother during pregnancy and malnutrition were important aetiological factors.

McKenzie and Craig (1955), in a pathological examination of a case, found absence of the stapedia artery associated with gross malformation of the maxillary artery. They suggested that a defect of the stapedia artery causes a maldevelopment not only in its own field of supply but also in the region of the first visceral arch, the vessels of which the stapedia artery normally supports during the critical phase between the disappearance of the first aortic arch and the full development of the external carotid. Weyers and Thier (1958) showed that rabbit embryos subjected to anoxaemia showed this type of defect.

François and Haustrate (1953), in an exhaustive review, classified a number of cases having the common feature of aplasia of the lower maxillary bone into a group which they termed the "syndrome of the first arch", in which they included mandibular dysostosis, mandibulo-facial dysostosis, the Streiff-Hallerman syndrome, and otocephaly. It seems clear that many related cases characterized by hypoplasia of the maxilla result from a defect in development of the first branchial arch. It is probable that this defect is associated with developmental anomalies of the maxillary artery resulting from an absence of the stapedia artery. Whether this is genetically determined or is the result of external factors is not certain.

It is thus possible to put forward explanations for the abnormalities found in encanthoschisis. Developmentally the caruncle is part of the margin of the lower lid which has become cut off by the development of the inferior canaliculus. There seem to be two possible explanations for the division of the caruncle into halves in cases of mandibulo-facial dysostosis. It may be that, if primarily genetically determined, the disturbance of the organization centre allows the caruncle to be separated off twice, once by an abortive development and secondly by the development of the true inferior canaliculus, thus giving the appearance of two caruncles.

However, if the absence of the stapedia artery is the primary cause of the Franceschetti syndrome, then one can suppose that the maxillary region will be poorly supplied with developing blood vessels, while the orbit, which is supplied by the ophthalmic artery via the internal carotid, will have a normal blood supply. It is possible that an abnormal anastomotic branch develops across the region of the inner canthus causing a division of the developing caruncle into two parts. When this small vessel regresses, a small band of fibrous tissue will be left passing between the two halves of the caruncle, leaving the picture of encanthoschisis.

Summary

Encanthoschisis comprises a group of abnormalities occurring at the inner canthus, which was first described by Hirsch (1912). It is typified by a split caruncle divided by a band of fibrous tissue, a horse-shoe shaped aperture of the inner canthus, and a defect in the plica semilunaris.

The syndrome, which should be known as Hirsch's syndrome, usually occurs in association with mandibulo-facial dysostosis or Franceschetti's syndrome.

A further case of encanthoschisis is reported, and a possible aetiology suggested.

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