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The proboscis in human cyclopia: an anatomical study in two dimensions

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ABSTRACT

A 2-dimensional anatomical study has been undertaken of the proboscis and its contribution to the roof of the median orbit in human cyclopia. The cyclops material consists of 4 sectioned fetal heads and a dried cyclops skull. The skeleton of the proboscis is formed by the nasal capsule. The base of the proboscis lies in the floor of the anterior cranial fossa filling an extended ethmoidal notch and contributing to the roof of the median orbit anterior to the fused lesser wings of sphenoid. The cavity of the proboscis is lined with squamous epithelium, respiratory and olfactory mucosa. Olfactory fibres pass from the proboscis into the extradural space of the ethmoidal notch forming a collection of tissue similar to the inferior layer of the normal olfactory bulb. The data indicate that the proboscis represents the anterosuperior part of the normal nasal cavity developed in the absence of median components. It is suggested that the cyclops face constitutes a model for the study of the development of the normal face.

INTRODUCTION

The proboscis presenting as a skin-covered tubular appendage above the median eye is a feature of many cases of human cyclopia. Brief descriptions of the proboscis have been included in reports on cyclops specimens by a number of authors (Deligdisch et al. 1978; Kokich et al. 1982). A more detailed account has been given by Torczynski et al. (1977). The roof of the median orbit has been described as being formed by the orbital plates of the frontal bone meeting in the median plane (Sarma, 1963). Other authors have described extension of the cavity of the proboscis towards the floor of the anterior cranial fossa (Keith, 1948; Duke-Elder, 1964) and Mayou (1906) reported the presence of an olfactory nerve in 1 case of human cyclopia. The present paper presents data on the proboscis and its contribution to the roof of the median orbit based on the study of a dried cyclops skull and of 4 cyclops heads sectioned in the sagittal and coronal planes. Data arising from this study and from earlier studies on the upper jaw and floor of the median orbit of the same specimens may be of value in the study of the development of the normal face.

MATERIALS AND METHODS

The perinatal material and the methods used in its study have been described previously (McGrath, 1989*a*). Two heads, Cyclops 1, 4, and 2 normal heads were sectioned in the sagittal plane. Two heads, Cyclops 2, 3, and 1 normal head were sectioned in the coronal plane. All sections were cut at 15 μm. Most sections were stained with PAS-orange G. The coronal sections presented in Figures 6–11 are viewed from the anterior aspect. A dried cyclops skull and 4 normal perinatal skulls were used in the study. As in previous studies of this cyclops material (McGrath, 1989*a*, *b*; McGrath and Sperber, 1990) the data are based mainly on the well preserved Cyclops 1 and 2. The brain tissue was not suitable for detailed examination in any cyclops specimen. To facilitate correlation between the figures of the dried skull and those of sectioned material in the sagittal and coronal planes, reference levels are indicated in Figures 2 and 5.

RESULTS

The proboscis as an external facial feature in cyclopia is shown in Figure 1. The skin-covered appendage is

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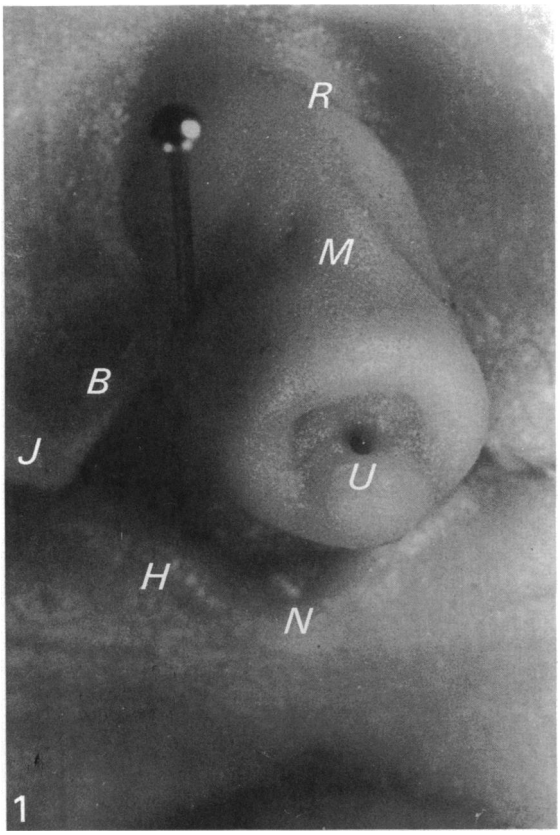

cyclindrical in shape. Its root is fixed. It is of the order of 10 mm long with a maximum external diameter of 10 mm. The appendage has a single aperture. The relationship of the eyelids to the appendage gives the impression that the proboscis is situated above the median orbit. However, in the dried cyclops skull the remnant of the proboscis is seen filling the ethmoidal notch in the roof of the orbit. The ethmoidal notch is bounded anteriorly by the superior orbital margin, laterally by the orbital plates of the frontal bone and posteriorly by the lesser wings of sphenoid which are fused around a median optic foramen (Fig. 2).

Sections through the proboscis are seen in sagittal section in Figures 3–5, Cyclops 1 and in coronal section posterior to anterior, in Figures 6–11, Cyclops 2. To facilitate presentation of the findings structures which appear to correspond with structures in the normal material are identified as such.

Fig. 1. Proboscis, external view, Cyclops 1. The proboscis presents as a tubular appendage with a fixed root (*R*), mobile distal part (*M*) and single aperture (*U*). A pin placed to the right of the proboscis to maintain alignment between root and distal part has drawn the lacrimal part (*B*) of the right upper eyelid superiorly. *N*, median caruncle on floor of orbit; *H*, lacrimal part of lower eyelid; *J*, lacrimal papilla of right upper eyelid.

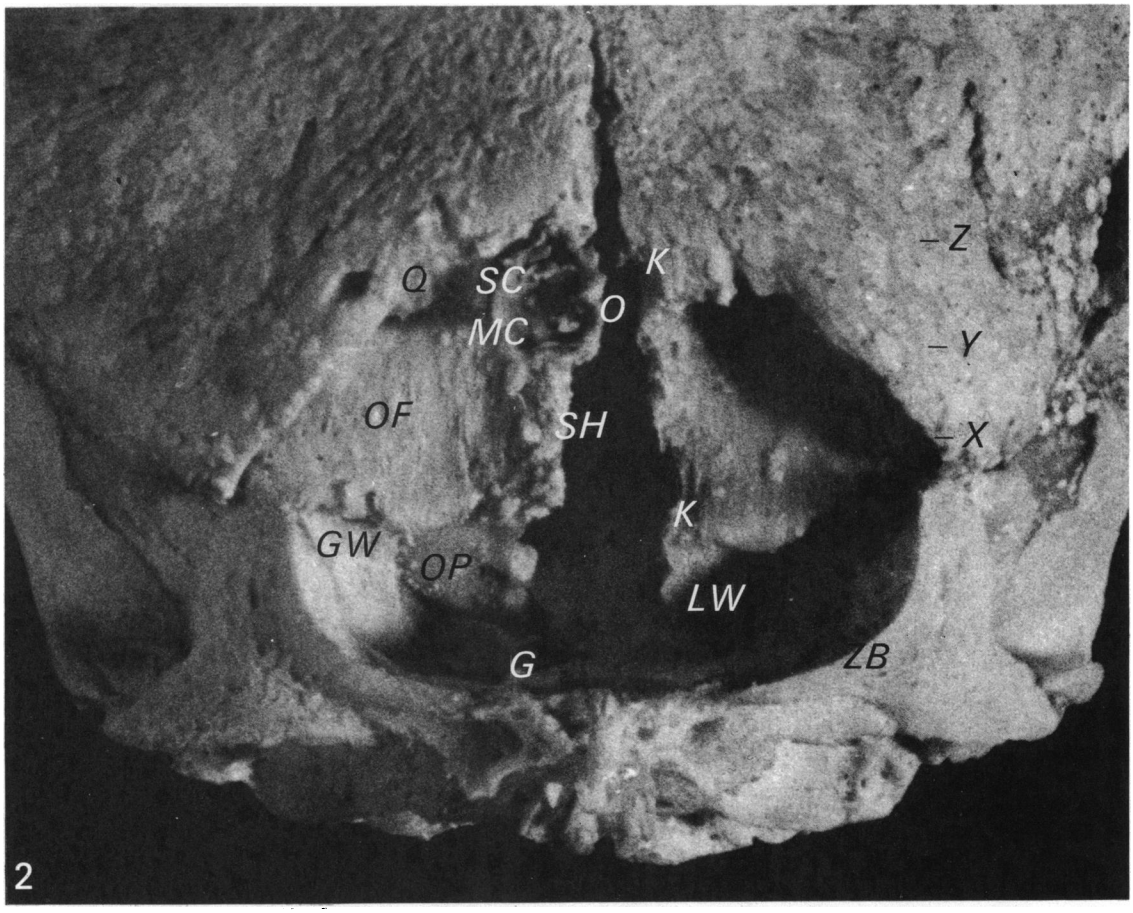

Fig. 2. For legend see opposite.

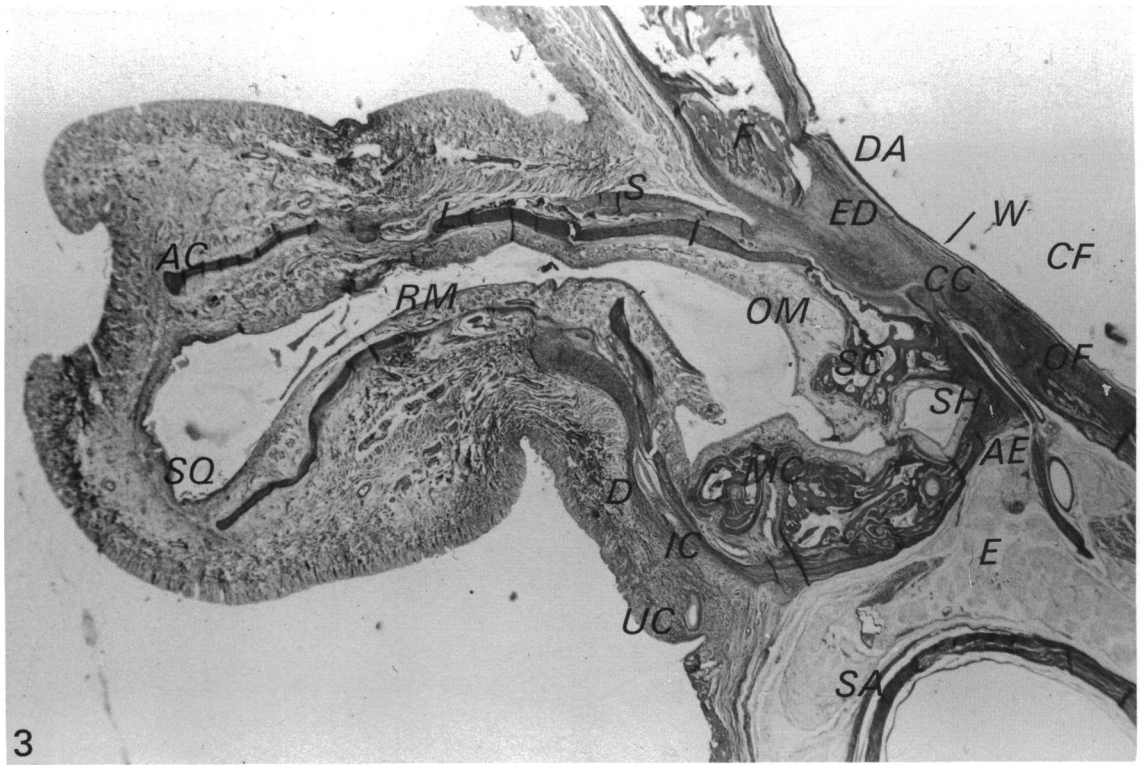

Fig. 3. Sagittal section through proboscis just within left edge of ethmoidal notch. Cyclops 1. The proboscis presents in 3 parts: the base in the ethmoidal notch, the labyrinth as an orbital mass, and distally in the root of the external appendage, and the mobile part of the external appendage. Anterior extremity of ethmoidal notch at level W; posterior extremity at anterior edge of lesser wing of sphenoid (unlabelled) at right margin. *CF*, anterior cranial fossa; *DA*, dura-arachnoid; *ED*, extradural fibrous tissue, *F*, frontal bone; *CC*, cranial part of cartilage of base of proboscis; *OF*, edge of orbital plate of left frontal bone; *AE*, anterior ethmoidal nerve and artery; *IC*, inferior concha; *OM*, olfactory mucosa; *I*, persistent cartilage of nasal capsule; *S*, fibrous sheath; *L*, lateral nasal cartilage; *RM*, respiratory mucosa; *AC*, major alar cartilage; *SQ*, squamous epithelium; *UC*, superior canaliculus; *D*, nasolacrimal duct; *E*, orbital fat; *SA*, sclera. ×5.5.

As seen in section the proboscis presents as a fibrocartilaginous structure roughly cylindrical in shape. It encloses a cavity which has a single, external, aperture. The proboscis presents in 3 contiguous parts – the base which fills the ethmoidal notch, the labyrinth which projects into the orbit and beyond the orbital margin forms the fixed root of the external appendage, and the distal part of the external appendage. The cavity enclosed within the proboscis extends from the external aperture some 18 mm to the base. The cavity of the external appendage is an open passageway of some 4 mm in diameter. More proximally the passageway occupies the superior part of an expanded cavity. The inferior part of the expanded cavity is almost filled with osseous protrusions (Figs 3, 4, 8–11).

The base of the proboscis is formed by an irregular mass of cartilage which, together with extradural fibrous tissue and neurovascular elements, fills the ethmoidal notch contributing both to the floor of the anterior cranial fossa and to the roof of the median orbit (Figs 2–8). The dimensions of the base are of the order of 6 mm × 3 mm × 1.5 mm. On its orbital aspect the base presents as a cribriform plate anteriorly (Fig. 8) and as a rod of cartilage posteriorly (Figs 4, 6). The cavity of the proboscis extends into this posterior part of the base (Figs 4, 5, 7). The rod of cartilage is identified as the sphenoidal conchal element and the cavity as a median sphenoidal sinus. On the cranial aspect of the base are 2 parasagittal ridges of cartilage. Each ridge is closely applied to the medial edge of the orbital plate of the frontal bone (Figs 6–8). The

Fig. 2. Dried cyclops skull, median orbit presenting. Distraction line passes through the superior orbital margin (*Q*), then close to the left edge of the ethmoidal notch (*K–K*) and through the roots of the left lesser wing of sphenoid (*LW*). In the anterior part of the ethmoidal notch the orbital plates of ethmoid (*O*), superior conchae (*SC*) and middle conchae (*MC*) obscure the cribriform plate. More posteriorly the ethmoidal notch is filled by the sphenoidal conchal element (*SH*). The rough appearance of the region of the ethmoidal notch is in contrast with the smooth surface of the orbital plate of frontal bone (*OF*) and of the lesser wings of sphenoid. *OP*, median optic foramen; *GW*, greater wing of sphenoid; *ZB*, zygomatic bone; *MX*, maxilla, *G*, median lacrimal fossa in floor of median orbit. Reference levels: *Z*, *Y*, *X*, Figs 5–8. ×3.7. In succeeding figures, only additional abbreviations are given.

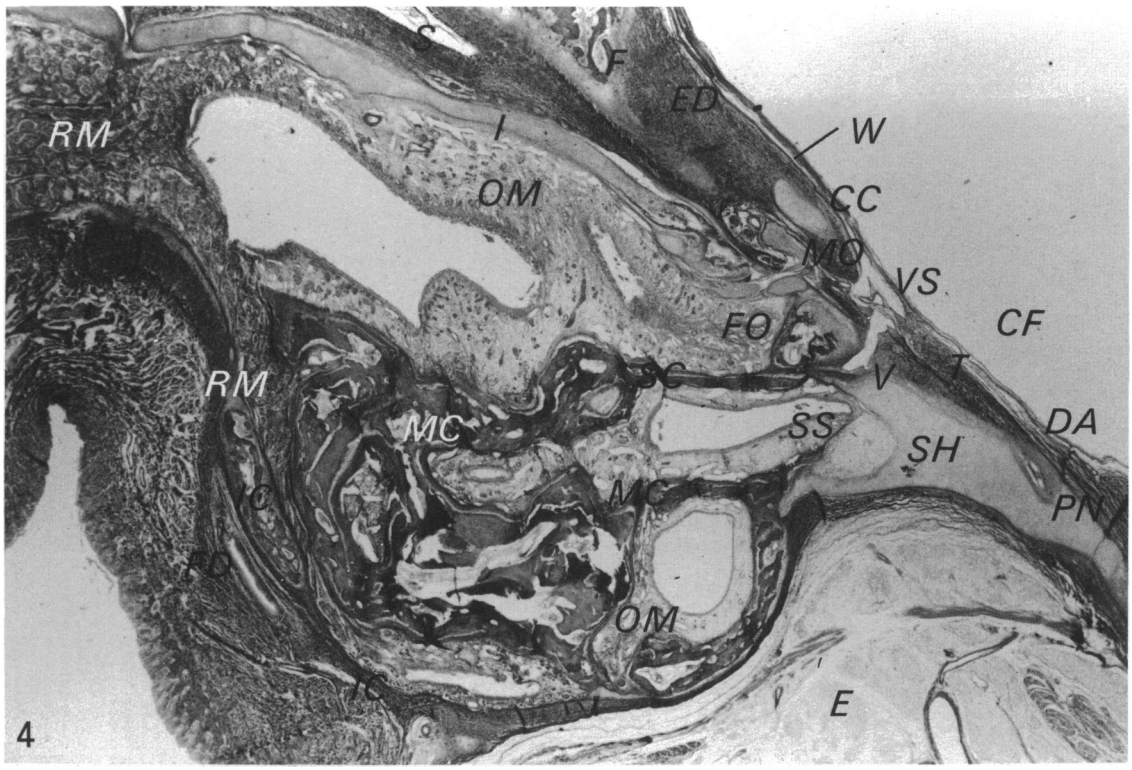

Fig. 4. Sagittal section through proboscis close to median plane, Cyclops 1. Olfactory fibres (*FO*) pass superiorly through the cribriform plate (unlabelled). Respiratory mucosa extends proximally into the labyrinth covering the internal surface of the inferior conchae. Fused nasolacrimal ducts (*FD*) lie in close contact with the external surface of the inferior conchae. MO, mass of olfactory fibres; *VS*, venous sinuses; *V*, vein; *T*, band of olfactory fibres; *SS*, sphenoidal sinus; *PN*, posterior branch of nasociliary nerve. × 10.

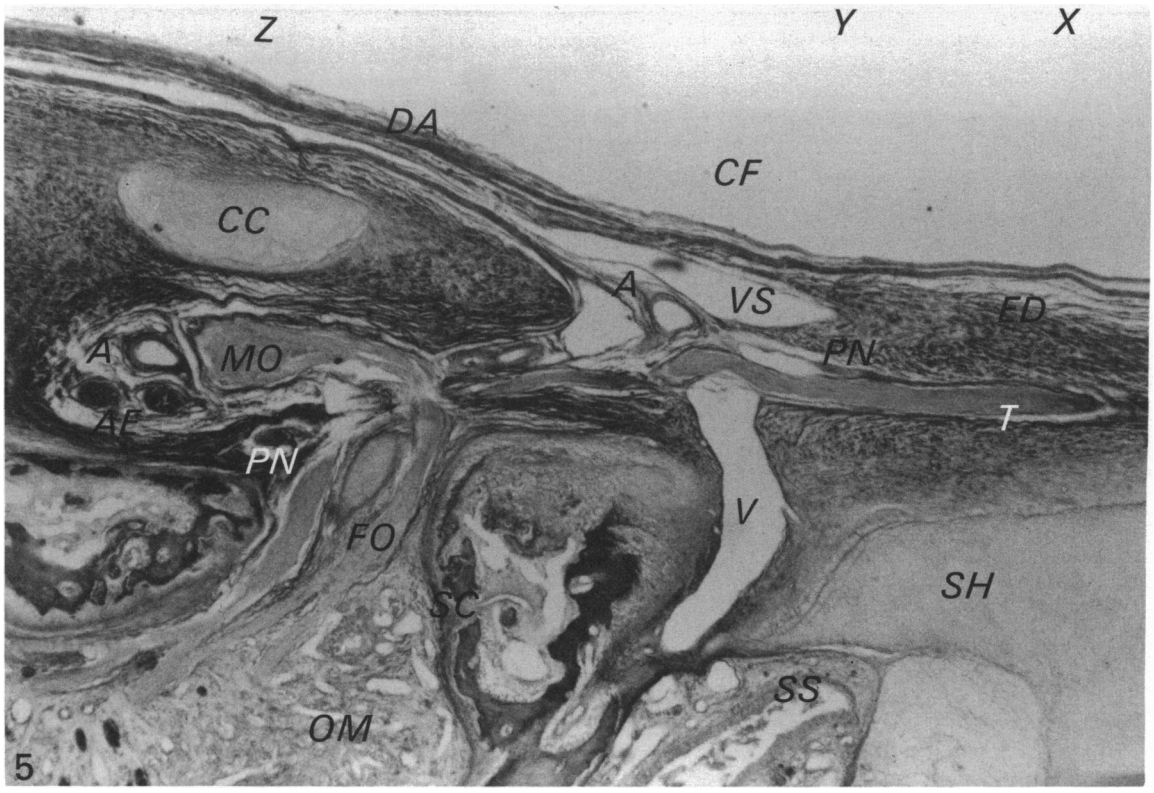

Fig. 5. Sagittal section through proximal proboscis close to median plane, Cyclops 1. The section has been rotated anticlockwise through 45°. Olfactory fibres passing through the cribriform plate (unlabelled), arch towards the olfactory mass anteriorly and into a band of fibres which extends posteriorly embedded in extradural fibrous tissue. The 2 anterior ethmoidal nerves lie anteroinferior to the olfactory mass. In this specimen these nerves continue as the external nasal nerves. *A*, artery. × 35.

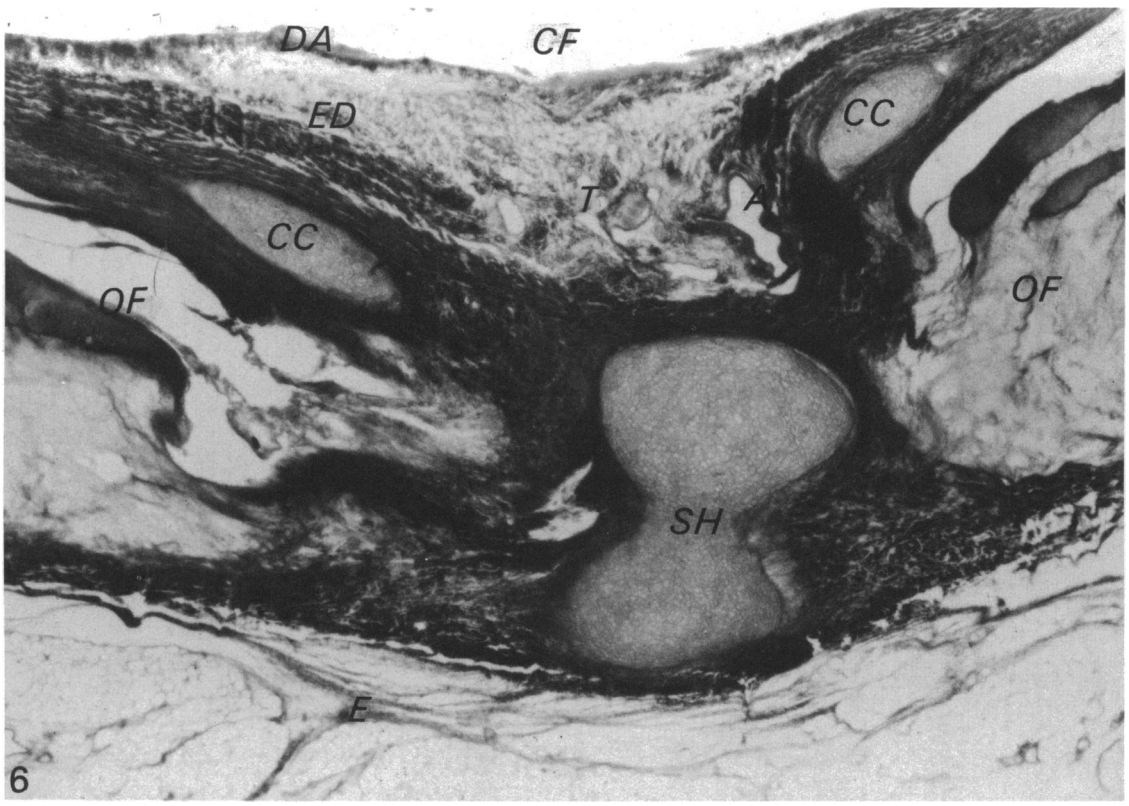

Fig. 6. Coronal section through posterior part of base of proboscis within ethmoidal notch, Cyclops 2 (level X, Figs 2, 5). The orbital part of the base presents as a median cartilaginous mass identified as the sphenoidal conchal element. The cranial part presents on each side as cartilage applied to the medial edge of the orbital plate of the frontal bone. A small collection of palely staining fibres (*T*) may be the posterior tip of a median band of olfactory fibres (Fig. 5). The cranial branch (*A*) of the left ophthalmic artery is within the ethmoidal notch. ×35.

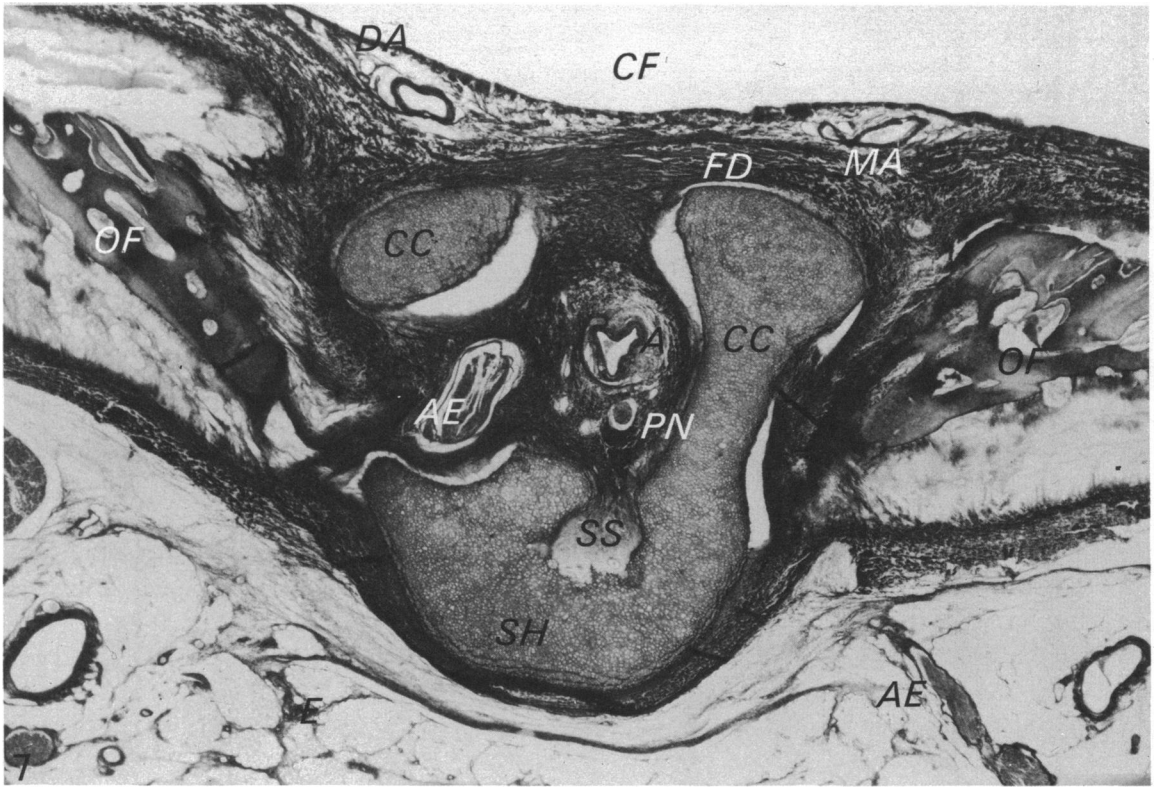

Fig. 7. Coronal section through base of proboscis, Cyclops 2 (level Y, Figs 2, 5). The cranial branch of the ophthalmic artery (Fig. 6) is now median in position. It is surrounded by a plexus of palely staining nerve fibres and veins (unlabelled). *MA*, meningeal branch of median artery. ×20.

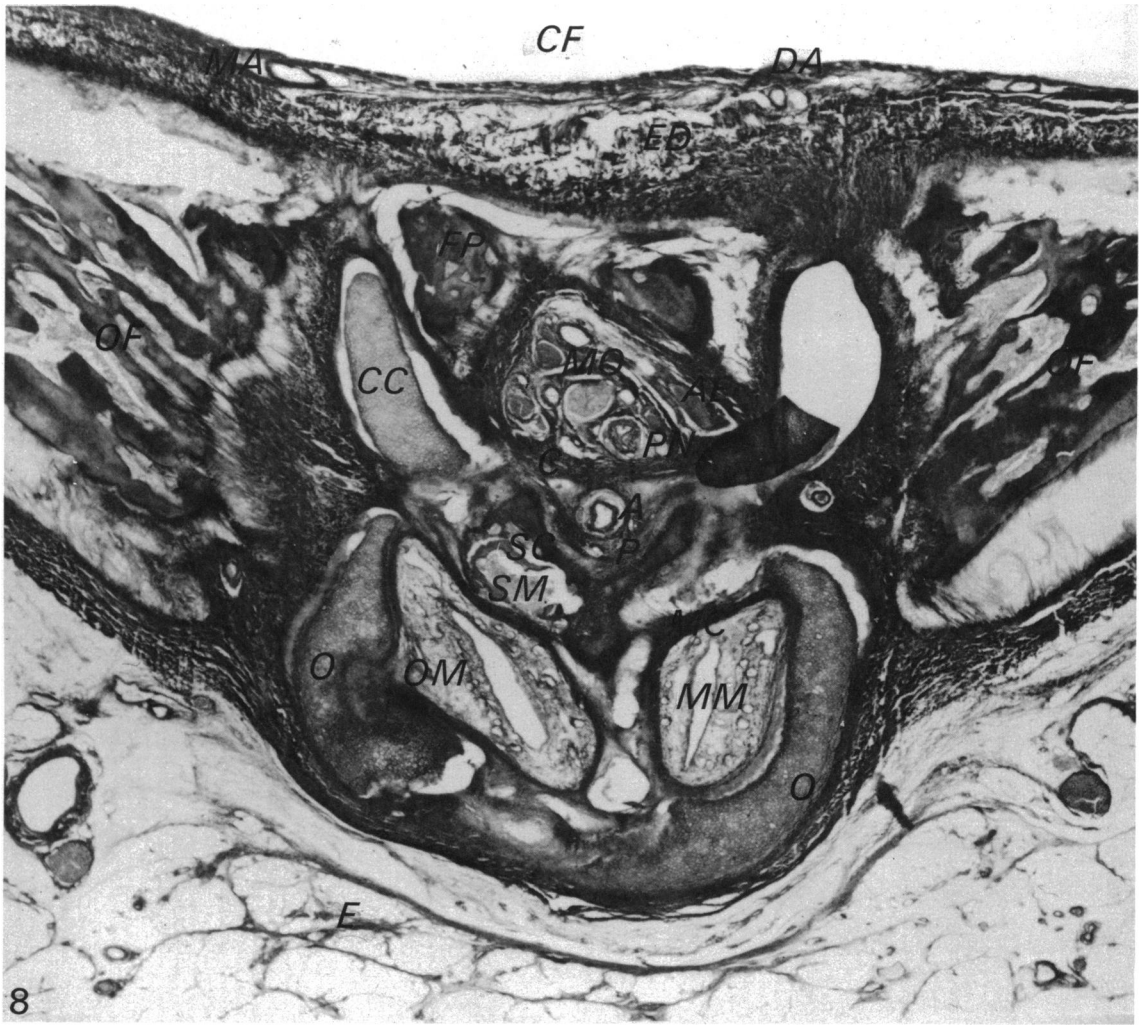

Fig. 8. Coronal section through anterior part of ethmoidal notch, Cyclops 2 (level Z, Figs 2, 5). The base and labyrinth of the proboscis are defined by the median cribriform plate (*C*). A mass of olfactory fibres presents as a palely staining median structure with an inferior tail. The right anterior ethmoidal nerve has divided into external and internal nasal nerves (unlabelled). Ossified elements (*FP*) are the posteriorly projecting edge of the frontal boundary of the foramen caecum. The lumen of the main passageway (*P*) of the labyrinth is compressed. *SM*, superior meatus; *MM*, middle meatus. ×20.

anterior edge of each ridge contributes to the boundary of the foramen caecum.

Fibrous tissue and neurovascular elements fill the irregularities on the cranial aspect of the base of the proboscis. The neurovascular elements form a median bundle in the extradural space related inferiorly and laterally to the cartilaginous elements of the base (Figs 5–8). Venous sinuses are seen in the central part of the ethmoidal notch. These sinuses communicate with meningeal veins and with veins passing superiorly through the cribriform plate (Figs 4, 5). Branches of the ophthalmic arteries enter the ethmoidal notch between the cranial and orbital parts of the base (Figs 6, 7). They give rise to branches to the meninges. Other branches form part of the median neurovascular bundle and accompany the branches of nerves as described below (Figs 7, 8). Bands of palely staining fibres pass superiorly through the cribriform plate (Figs 5, 8). In sagittal section these fibres are seen to orientate towards the venous sinuses in the central part of the ethmoidal notch. Some fibres then arch anteriorly to a well defined palely staining fibrous mass. Others pass posteriorly to form a median band embedded in extradural fibrous tissue (Fig. 5). The dimensions of the anterior mass are of the order of 0.8 mm × 0.3 mm × 0.6 mm. The fibres within the mass present a whorled appearance. The full extent of the posterior band cannot be definitely determined in Cyclops 2, coronal, due to the presence of a median artery together with its perivascular sympathetic network. However, a median collection of palely staining fibres is present posterior to the point where the artery takes up its median position and this may be taken as the terminal part of the posterior band

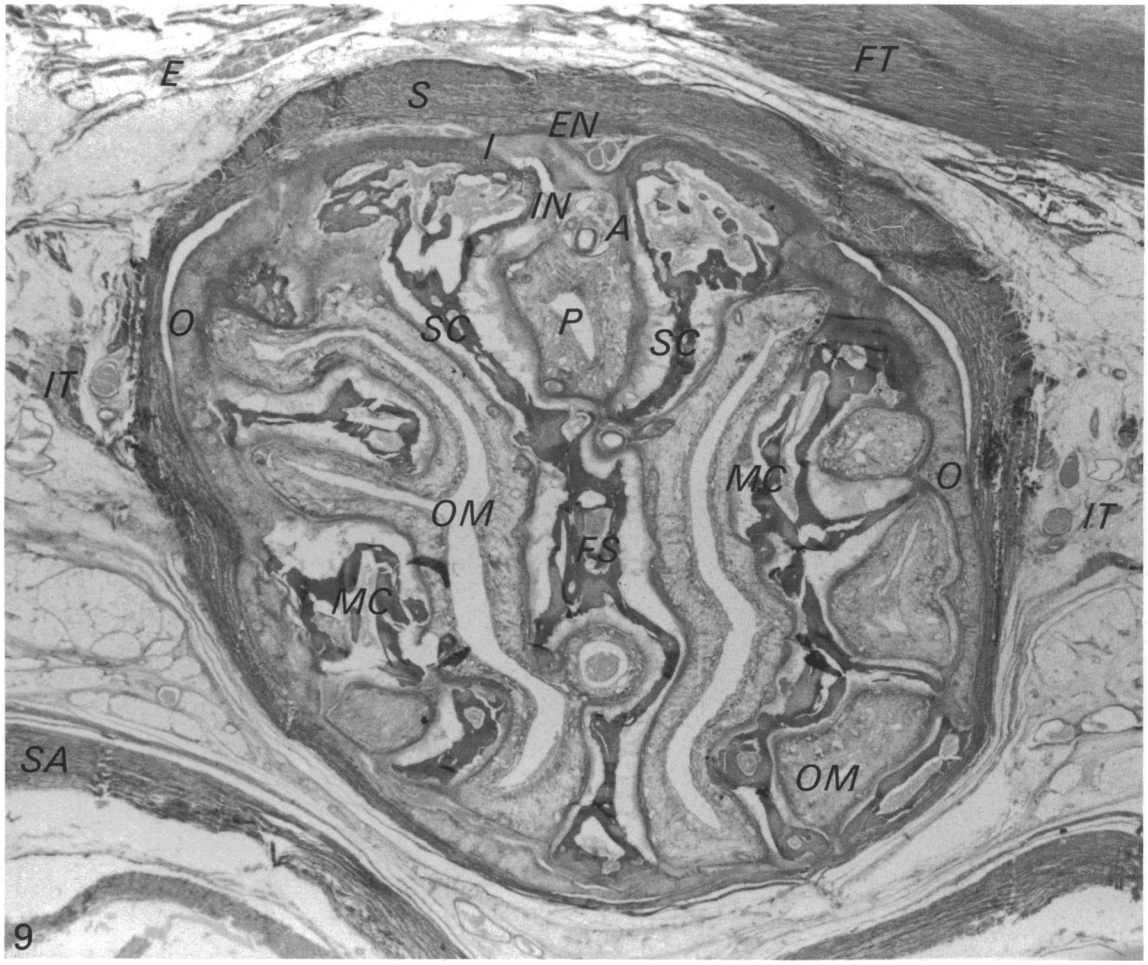

Fig. 9. Coronal section through labyrinth of proboscis close to anterior extremity of superior conchae, Cyclops 2. The narrow main passageway of the proximal labyrinth is supported by the superior surfaces of the superior conchae. The conchae fuse in the median plane (*FS*). *FT*, fibrous tissue underlying superior orbital margin; *IT*, infratrochlear nerve and artery; *EN*, external nasal nerves; *IN*, internal nasal nerves. × 15.

(Fig. 6). The dimensions of the posterior band are of the order of 2.4 mm × 0.12 mm × 0.12 mm. These neural elements are identified as olfactory fibres. No fibres could be found passing from these olfactory elements into the subarachnoid space of the anterior cranial cavity. The nasociliary nerve accompanied by a branch of the ophthalmic artery enters the ethmoidal notch on each side between the orbital and cranial parts of the base (Figs 3, 7). As the anterior ethmoidal nerve it runs forward in the peripheral part of the neurovascular bundle (Fig. 8). It may divide into external and internal nasal nerves as seen on the right side in Cyclops 2 (Fig. 8) or it may continue as the external nasal nerve as in Cyclops 1 (Fig. 5) and on the left side in Cyclops 2 (Fig. 8). In Cyclops 1 and on the left side in Cyclops 2 a posterior branch of the nasociliary nerve accompanied by a branch of the ophthalmic artery enters the ethmoidal notch posteriorly. The nerve runs anteriorly within the median neurovascular bundle (Figs 4, 7). It passes inferior to the olfactory mass (Figs 5, 8) and continues as the internal nasal nerve.

The labyrinth of the proboscis presents as a partially ossified cartilaginous rim contiguous with the cartilage of the base. Osseous turbinates project from the rim into the enclosed cavity. The external dimensions of the labyrinth are of the order of 10 mm × 9 mm × 8 mm. The cartilage of the anterosuperior quadrant is continuous with the cribriform plate. The cartilage forming the lateral quadrants of the rim is identified as the orbital plates of the ethmoid. In the proximal part of the labyrinth the orbital plates fuse to form the inferior quadrant of the rim (Figs 8, 9). In the anteroinferior part of the labyrinth the orbital plates are overlaid in the inferior quadrant by partially ossified cartilage identified as the inferior conchae fused inferiorly (Fig. 10).

The osseous turbinates which project from the inner superolateral parts of the rim are the superior conchae. The conchae are convex medially. They fuse in the

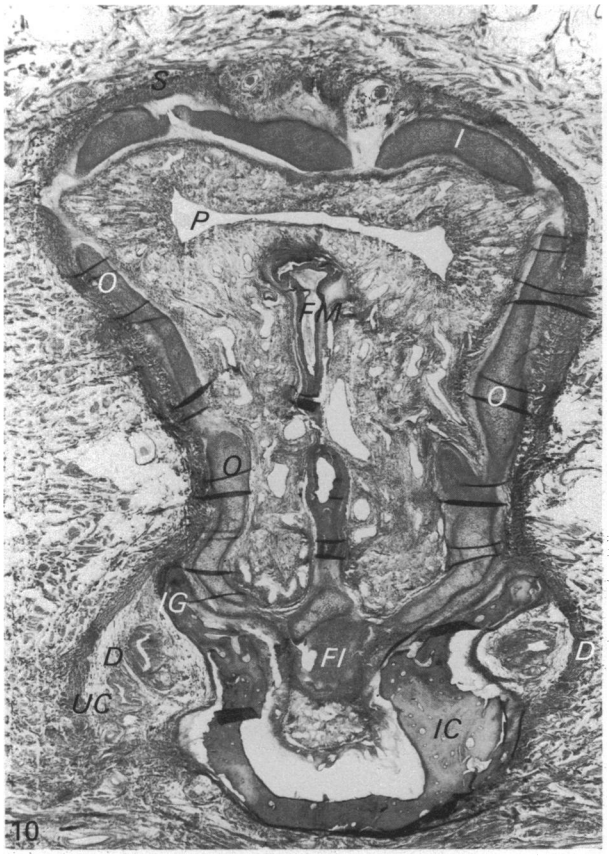

Fig. 10. Coronal section through labyrinth of proboscis, Cyclops 2. The section passes through the anterior part of the middle conchae and through the inferior conchae. A wide main passageway is supported by the superior surfaces of the middle conchae. The conchae fuse in the median plane (*FM*). The inferior conchae overlie the orbital plates laterally. Medially their convex internal surfaces fuse in the median plane (*FI*). A deep concavity on the external surface of each inferior concha accommodates a nasolacrimal duct. × 10.

median plane and extend inferiorly to the full depth of the cavity. Superiorly they form the floor of the narrow, main passageway (Figs 4, 8, 9). The anteroposterior extent of the superior conchae is of the order of 4 mm with a depth of 5 mm. The middle conchae present as a series of protuberances within the superior meatuses (Figs 3, 4, 8, 9). More distally the middle conchae meet in the median plane. They form the floor of the main passageway which in this section of the proboscis is about 4 mm wide (Figs 3, 4, 10). The anteroposterior extent of the middle conchae is of the order of 5.5 mm with a depth of 4 mm. In the anteroinferior part of the labyrinth the convex internal surfaces of the inferior conchae fuse in the median plane (Fig. 10). A median osseous rod formed by the fused inferior conchae projects anteriorly beyond the labyrinth (Fig. 11).

Part of the lacrimal drainage apparatus consisting of the upper canaliculi and the nasolacrimal ducts is associated with the external aspect of the labyrinth. The puncta of the upper eyelids are in line with those of the lower eyelids (Fig. 1). Each upper canaliculus is about 4 mm in length. It runs first medially (Fig. 11) and then vertically to the anterolateral aspect of the labyrinth where it opens into a nasolacrimal duct (Figs 3, 10). Each duct extends superolaterally about 1.5 mm, to end blindly, and inferomedially within a concavity on the external aspect of the inferior concha for some 2 mm to fuse with its fellow. The fusion forms a median multiloculated sac immediately subjacent to the most distal part of the labyrinth. The sac is supported by the median osseous rod formed by the fused inferior conchae (Figs 4, 11).

The nasal bones present as a fused osseous mass external to the cartilaginous rim immediately anterior to the superior orbital margin. The dimensions of the fused nasal bones are of the order of 1 mm × 0.5 mm × 0.5 mm. The external and internal nasal nerves and accompanying arteries pass from the anterior part of the base and run in close relationship to the cartilaginous rim of the labyrinth. The infratrochlear nerve arises as a branch of the nasociliary nerve and runs in close relationship to the lateral aspect of the labyrinth (Fig. 9).

The cartilage of the superior and lateral quadrants of the labyrinth extends distally to form the skeleton of the fixed root of the external appendage (Figs 3, 4, 11). The skeletal framework of the mobile part of the external appendage consists of segments of cartilage connected to the cartilage of the distal part of the labyrinth and to each other by fibrous tissue. The proximal segments of cartilage are thick and are identified as the lateral nasal cartilages. The distal cartilaginous segments are thin and fragmentary and are identified as the alar cartilages (Fig. 3). Between the fibrocartilaginous skeleton and the covering skin lie muscular and neurovascular elements which correspond to those present in the normal external nose (Figs 3, 11).

The cavity of the proboscis is lined with olfactory mucosa in the proximal labyrinth, respiratory mucosa over the inferior conchae and in the proximal part of the external appendage and squamous epithelium distally (Figs 3, 4, 8–11).

The skeletal framework of the proboscis contributes to the roof of the median orbit. The elongated ethmoidal notch is filled by the cribriform plate and sphenoidal conchal element which form the base of the proboscis. The proximal labyrinth suspended from the base corresponds to the ethmoidal segment of the normal interorbital mass.

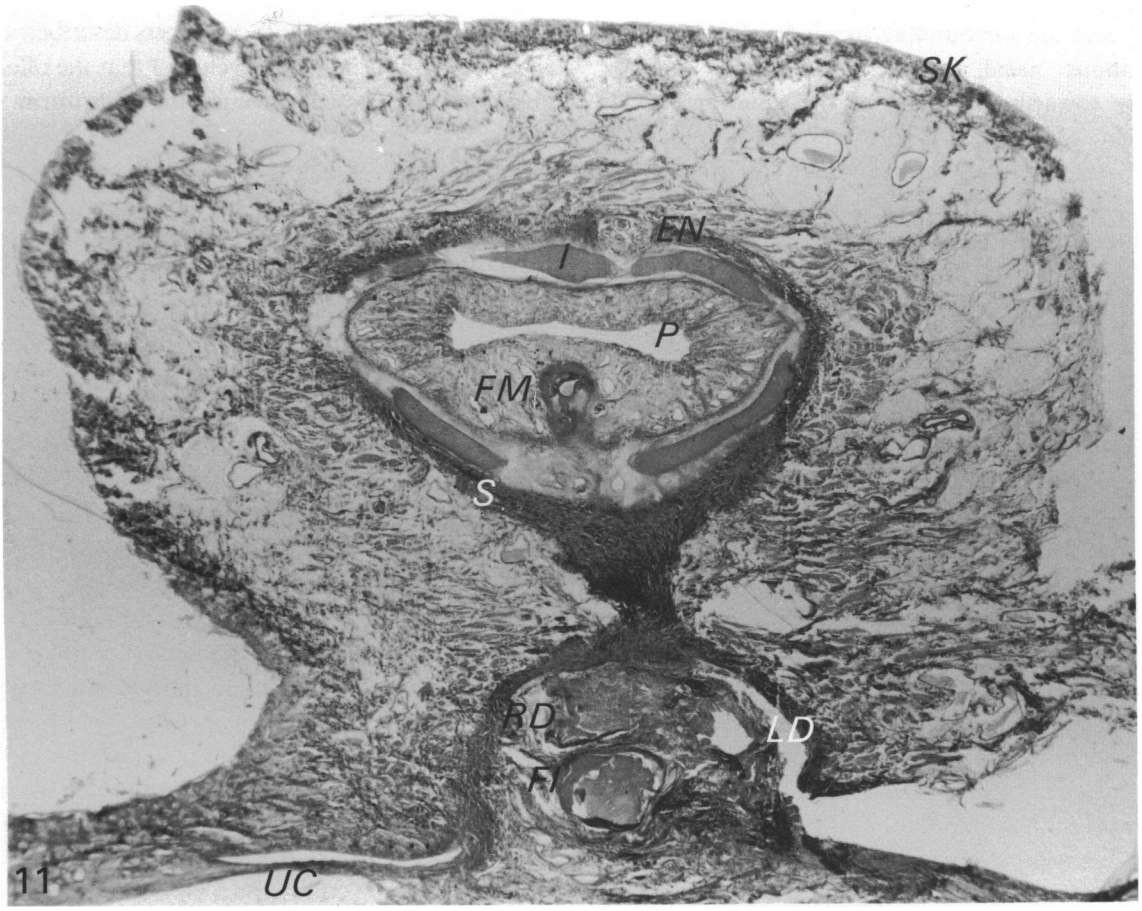

Fig. 11. Coronal section through fixed root of external appendage, Cyclops 2. The section passes through the distal labyrinth close to its junction with the mobile part of the external appendage. Inferiorly the section passes through the anterior tip of an osseous rod formed by the inferior conchae. *SK*, skin surface; *RD*, anterior wall of right nasolacrimal duct; *LD*, terminal part of left nasolacrimal duct. × 10.

DISCUSSION

Data on the proboscis included in general descriptions of cyclops specimens are limited in scope. A study of the proboscis and its contribution to the roof of the median orbit based on a comparable range of material and plane of section has not been previously presented. The current data indicate that the proboscis in human cyclopia approximates to the anterosuperior part of the nasal cavity developed in the absence of median components.

The major structures associated with the normal nasal cavity which are absent in cyclopia are the presphenoid, perpendicular plate of ethmoid, crista galli, septal cartilage and the anterior part of vomer. The medial crus of each major alar cartilage is also absent. The influence of median elements in the development of the nasal cavity has been considered by a number of authors. Moss et al. (1968) found that nasal and midfacial skeletal growth are independent of each other. Latham (1971) reported that the cyclopean maxilla showed a normal growth pattern. Bergland and Borchgrevink (1974) reported that the nasal septum cannot play any influential role in midfacial growth in man. McCann et al. (1991) found that in the chick the frontonasal process plays a major role in midfacial morphogenesis. The particular significance of the presphenoid in the development of the nasal cavity had not been reported. In cyclopia the presphenoid and the contiguous elements of the median segment of the anterior skull base are absent. The overlying brain presents as alobar holoprosencephaly. There is a single optic nerve. However, the structures in the skull base which are present and the associated soft tissues appear to achieve as normal a development as the absence of median structures permits (McGrath, 1989*a*, *b*; McGrath & Sperber, 1990).

While the morphogenesis of cyclopia has yet to be established the findings of the present study may support the following sequence in terms of the development of the proboscis. A median olfactory placode develops which comes to be surrounded by the lateral nasal processes. The placode deepens into

a cavity and the surrounding tissue develops into the cartilaginous nasal capsule. The cavity extends cranially towards the floor of the anterior cranial cavity. In the absence of the presphenoid, posteroinferior deflection of the expanding cavity towards the developing nasopharynx does not occur. The nasal capsule related to the posterosuperior part of the cavity develops into a sphenoidal conchal element which fills the posterior part of the elongated ethmoidal notch. The nasal capsule related to the superior part of the cavity develops into a cribriform plate which fills the anterior part of the ethmoidal notch. On the cranial aspect of the cartilage within the ethmoidal notch a lateral ridge is present closely associated with the medial edge of the orbital plate of the frontal bone. A similar relationship is seen in normal material and this finding might indicate that the cartilaginous nasal capsule contributes to the roof of the ethmoidal air sinuses. The anterior edge of each ridge has the same relationship to the foramen caecum as the normal alar process of the crista galli.

The cartilaginous nasal capsule related to the anterosuperior quadrant of the cavity and lying immediately inferior to the nasal bones persists as in normal material. The nasal capsule related to the lateral aspects of the proximal cavity develops into the orbital plates of the ethmoid which support the superior and middle conchae. Posteriorly the orbital plate is contiguous with the sphenoidal conchal element. Anteriorly the cartilage extends into the root of the external appendage. The general features of these ethmoidal elements approximate to those of the normal facial ethmoid. The inferior concha develops in the anteroinferior part of the nasal capsule. Its external surface is related to the nasolacrimal duct as in the normal nasolacrimal canal. There is no contact with the maxilla, lacrimal and palatine bones which in cyclopia are in the floor of the median orbit. The convex inner surface of the inferior concha represents the turbinate. In the distal mobile part of the proboscis the parasagittal structural features are similar to those of the normal nose. In the absence of a nasal septum tethered to the upper jaw, the distal part of the proboscis presents as a freely mobile facial appendage.

The general features of the cavity of the proboscis are similar to those of the anterosuperior part of the nasal cavity. There are no inferior meatuses, the nasolacrimal ducts being related to the inferior conchae external to the cavity of the proboscis.

The venous sinuses which are present on the cranial aspect of the base in Cyclops 1 receive a large vein draining through the cribriform plate. A large ethmoidal vein draining into the venous plexuses around the normal olfactory bulb has been described (Durward, 1964). It is noted in Cyclops 1 that the olfactory fibres having passed through the cribriform plate converge on the site of the venous sinuses. It might be suggested that the venous sinuses represent the normal position of the olfactory bulb and that the olfactory fibres having failed to make a connection with the brain in this site become embedded in adjacent extradural fibrous tissue. The whorled appearance of the fibres forming the anterior mass is similar to that of the inferior layer of the olfactory bulb.

It is evident from the present study that in cyclopia the lacrimal drainage apparatus develops as isolated upper and lower components. These data also indicate that the earlier identification of the nasolacrimal duct in the lower component of the lacrimal drainage apparatus is incorrect (McGrath & Sperber, 1990).

The contribution made by the proboscis to the roof of the median orbit is the cartilaginous base which fills the elongated ethmoidal notch. The base suspends the proximal labyrinth which approximates to the ethmoidal component of the normal interorbital mass. This normal interorbital mass is completed posteriorly by the presphenoid, absent in cyclopia, and anteriorly by the lacrimal bone and the lateral part of the frontal process of the maxilla which in cyclopia are in the floor of the median orbit.

Data arising from the present study and from earlier studies on the upper jaw and floor of the median orbit indicate that in cyclopia the face is as normal as the absence of median structures permits. It is suggested that the face in cyclopia, in both its positive and negative aspects, constitutes a model for the study of the development of the normal face.

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