Congenitally malformed cystic eye, causing extensive protrusion of upper eyelid, and complete extrusion of conjunctival sac through the palpebral fissure / by S. Johnson Taylor and E. Treacher Collins.

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Congenitally malformed cystic eye, causing extensive protrusion of upper eyelid, and complete extrusion of conjunctival sac through the palpebral fissure.

By S. Johnson Taylor and E. Treacher Collins.

Clinical notes by S. Johnson Taylor.

The subject of this communication, J. E. W—, an illegitimate male child, æt. 3 weeks when first seen, was brought to me from the country early in June, 1904, with a large cystic protrusion occupying the position of the left eye, and apparently mainly formed by a bulging forwards of the conjunctiva of the upper lid; noticed at the birth of the child, it had increased since; it was the only deformity about the little patient, and the right eye was quite normal.

The infant was too young to stand the removal of the mass, so, from an exploratory and temporising point of view, I at once tapped it with a fine trochar and cannula, and removed a considerable quantity of light straw-coloured fluid. The cyst then collapsed and some rather solid tissue, which I imagined represented the eye, could be felt in the orbit with the finger.

Nearly a year elapsed before I saw the child again; the cyst refilled the day after it was tapped, and had gone on increasing in size until it projected 2½ inches from the orbital rim, and looked very much like a large, paraphimosed penis; for some two months it had been discharging and bleeding occasionally, and was pulling the little patient down. Fig. 1 fairly well represents its appearance and proportions, the part projecting forwards from the lids being covered with mucous membrane, which seemed to be mainly the conjunctiva of the upper lid, slight constriction being caused by the margins of the lids.

The child was now a year old, and the condition was telling on its health, so I had no hesitation in advising its

removal, my object being, if possible, to get it out *en masse*.

This I succeeded in doing by making an incision just through the skin all round close to the margins of the lids, then burrowing under and dissecting up the skin flaps until I could pass a pair of curved scissors well behind, and so separating all the connections, not a drop of the contents escaping.

The child was very collapsed after the operation, and remained in a doubtful condition for some days, but ulti-

Fig. 1.

Shows the appearances of the cystic protrusion shortly before operation.

mately recovered; and when seen last week was fat and well, the cut margins of the upper and lower lids having completely united.

Pathological report by E. Treacher Collins.

The specimen consists of the contents of the right orbit, including the whole conjunctival sac and the margins of the eyelids.

The whole of the conjunctiva, palpebral, ocular, and retrotarsal folds is everted through the palpebral aperture, forming a prominent, cystic, rounded swelling. The surface of the conjunctiva is everywhere thrown into rugæ, and is covered with papillæ.

It is difficult to differentiate the surface of the cornea from the conjunctiva; there is, however, a little below the middle line of the protuberant mass a circular area 9 mm. in width, which is depressed slightly below the level of the surrounding tissue, from which it is separated by a somewhat lipped edge. The surface of this area is not papillated like the surrounding tissue, but it has what looks like a membrane covering it.

Above the margin of the upper lid, where the skin has been cut through, there is a large, thin-walled, collapsed cyst, which appears to be part of the other cystic protrusion extending through the palpebral aperture.

At the lower and posterior part of the specimen is a resistant structure presenting the appearance of normal sclerotic.

No optic nerve can be differentiated. The extra-ocular muscles are large, some are inserted into the piece of scleral-like tissue, and some into the walls of the cyst.

After the specimen had been divided into two by an antero-posterior vertical section, the upper and anterior part was seen to be entirely occupied by a collapsed cyst, the walls of which varied somewhat in thickness in different parts, being thinnest at the extreme upper part. A partition springing from the anterior and lower part of the outer wall partly divided the cavity of the cyst into two.

On the inner surface of the piece of sclerotic some pigmented tissue like that of normal choroid was seen. There was no pigmented tissue anywhere else in the wall of the specimen. The posterior part of the cavity of the cyst was encroached upon by a mass of solid tissue protruding from the lower outer wall. This mass of tissue presented a yellowish grey colour on section.

Microscopical examination. Margins of eyelids and conjunctiva.—Sections through the upper eyelid show the tarsus to have become doubled inwards on itself about its centre (Pl. VI, fig. 2). The tarsus in the lower lid is not bent to the same extent, but the conjunctiva of the retrotarsal fold commences to turn forwards directly where it ceases to be adherent to the tarsus.

Immediately beneath the epithelium of the skin of the upper lid there is much small round-cell inflammatory infiltration. Similar round-cell infiltration beneath the skin of the lower lid is confined to its free margin.

The conjunctiva of the upper retrotarsal fold, which is entirely everted, is covered with numerous papillæ having deep recesses between them. Immediately beneath the epithelium, especially in the vicinity of Krause's glands, there is much small round-cell infiltration, the ducts and loculi of the glands appearing imbedded in it.

The surface epithelium is several layers thick, the most superficial cells being flattened, but none scaly or devoid of nuclei; a few goblet-cells are here and there seen.

On passing away from the vicinity of Krause's glands the papillated condition of the surface of the conjunctiva continues, there is much new formation of lymphoid tissue beneath the epithelium, and much dilatation of lymphatic spaces.

Cornea.—Sections which pass through the circular depressed area, previously referred to as possibly representing the cornea, show that the epithelium which forms numerous papilla and downgrowths up to its margin, there ceases as a continuous layer. There is laminated fibrous tissue much infiltrated by leucocytes, very densely so at the anterior part, where clumps of epithelial cells imbedded in masses of round cells are to be seen. On the free surface of this infiltrated tissue is a layer of what appears to be some coagulated homogeneous substance.

Outer wall of cyst.—The piece of sclerotic at the lower and posterior part of the specimen presents the usual

histological appearances of that structure; it measures 7 mm. in length. Anteriorly it tapers off into a thin layer of fibrous tissue, with regularly arranged bundles of fibres, which composes the outer wall of the cyst. Posteriorly it ends somewhat abruptly in much the same way as the sclerotic nominally does, at the margin of the optic nerve. There is there a gap 6 mm. wide, filled mainly by atypical vitreous, to be afterwards described. On the further side of this gap a second piece of sclerotic 3 mm. in length is encountered, with a small nodule of typical hyaline cartilage imbedded in its anterior part (Pl. VII, fig. 4). This second piece of sclerotic also tapers off into the fibrous tissue forming the external wall of the cyst.

Throughout its whole extent the cyst has an outer fibrous tissue wall varying in thickness in different parts, and having here and there patches of round-cell infiltration in it. Anteriorly the conjunctiva is seen to be separated from the outer cyst wall by loose tissue and lymphatic spaces corresponding to Tenon's capsule. Over one area in the anterior part of the specimen, corresponding to that already described as probably cornea this differentiation of conjunctiva from the outer wall of the cyst is not to be made out. Here the outer wall of the cyst seems to come directly to the surface.

Middle wall of cyst.—Lining the piece of well-formed sclerotic at the lower and outer part of the specimen is a piece of choroid presenting quite its normal appearance, and having on its inner surface the elastic lamina and pigment epithelium. The stroma of this piece of choroid is devoid of pigment, but its vascular and lymphatic layers are well formed, and a ciliary nerve is seen in section of healthy appearance. Just previous to the anterior termination of the normal sclerotic this choroidal tissue passes into a mass of unstriated muscle-fibres resembling those of the ciliary muscle, and having on its inner surface projections like the ciliary processes.

Between the outer fibrous tissue covering of the cyst

and its inner wall of degenerate retinal tissue, to be described directly, there is tissue containing blood-vessels more or less widely spaced out, but devoid of any pigmented cells. This probably represents attenuated choroid.

Inner wall of cyst.—The inner lining tissue of the cyst varies in different parts of the specimen. Nowhere is there any retina with well differentiated layers, and nowhere can those parts of the retina, which are developed from the two layers of the secondary optic vesicle, be found lying in apposition.

On the inner surface of the portion of choroid which is well developed, and of the rudimentary ciliary processes in which it terminates, a layer of pigment epithelial cells presenting their normal appearance is seen. This layer forms part of the lining wall of the cyst, there being no retina on its inner surface.

Covering the greater portion of the solid mass of tissue which protrudes into the cavity of the cyst from below, and which, as will be mentioned later, is atypically developed vitreous humour, is a layer of epithelial cells. They resemble those of the outer layer of the retina, but are devoid of pigment, except for a few granules here and there. This slight tendency to pigmentation, and their continuity with the pigment epithelium already described, serve to show that they really represent the outer layer of the secondary optic vesicle.

The inner wall of the greater portion of the cyst, and of all its anterior part, consists of a degenerate kind of retina. It is composed of a close network of delicate fibres with branching cells and nuclear bodies, like those met with in the nuclear layers of the retina.

The partition proceeding backwards from the anterior wall of the cyst, and partially dividing its cavity into two parts (Pl. VI, fig. 1), is seen to be composed of two layers of membrane of this description, with fibrous tissue containing numerous blood-vessels between them, the latter being evidently of mesoblastic origin.

PLATE VI.

Illustrates Messrs. S. Johnson Taylor and E. Treacher Collins' paper on A Congenitally Malformed Cystic Eye causing Extensive Protrusion of the Upper Eyelid and Complete Extrusion of the Conjunctival Sac through the Palpebral Fissure (p. 177).

(For the microphotographs from which the figures on this plate are reproduced the writers are indebted to Mr. E. Collier Green.)

Fig. 1 shows a section through the whole everted conjunctival sac, the margins of the eyelids, and the cystic eyeball. (1) Margin of the upper eyelid, showing under higher magnification in fig. 2 on this plate. To the left of it is the conjunctiva everted through the palpebral aperture in front of lower part of cyst. To the right of it is the upper part of the cyst, which extended beneath the skin of the upper lid. (2) Cavity of cyst. (3) Malformed lens and lens capsule, shown under higher magnification in Plate VII, fig. 3. (4) A piece of well-formed sclerotic lined by well-formed choroid. (5) Double fold of retinal tissue extending back from anterior wall of cyst to the position of the lens. (6) Piece of sclerotic containing cartilage, shown under higher magnification in Plate VII, fig. 4. (7) Margin of lower eyelid.

Fig. 2 shows a section through the margin of the upper eyelid. The tarsus containing the Meibomian gland is seen to be doubled forwards on itself. On the right side is the thickened everted conjunctiva, with a papillated surface. On the left is the skin of the eyelid.

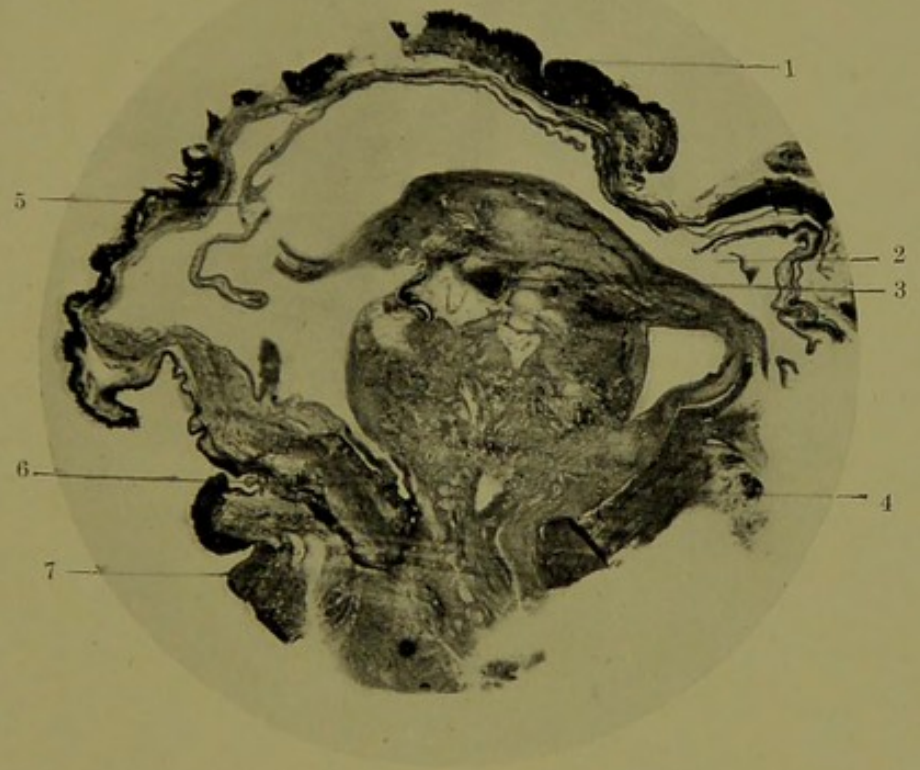

Fig. 1.

Fig. 2.

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PLATE VII.

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Fig. 3 shows a section through the malformed lens and its capsule. Anteriorly the capsule is thrown into numerous folds. Posteriorly there is a wide gap in it through which vascular mesoblastic tissue (atypically developed vitreous) has protruded and become mixed with epiblastic lens substance.

Fig. 4 shows a section through a piece of the sclerotic in which is imbedded a nodule of hyaline cartilage.

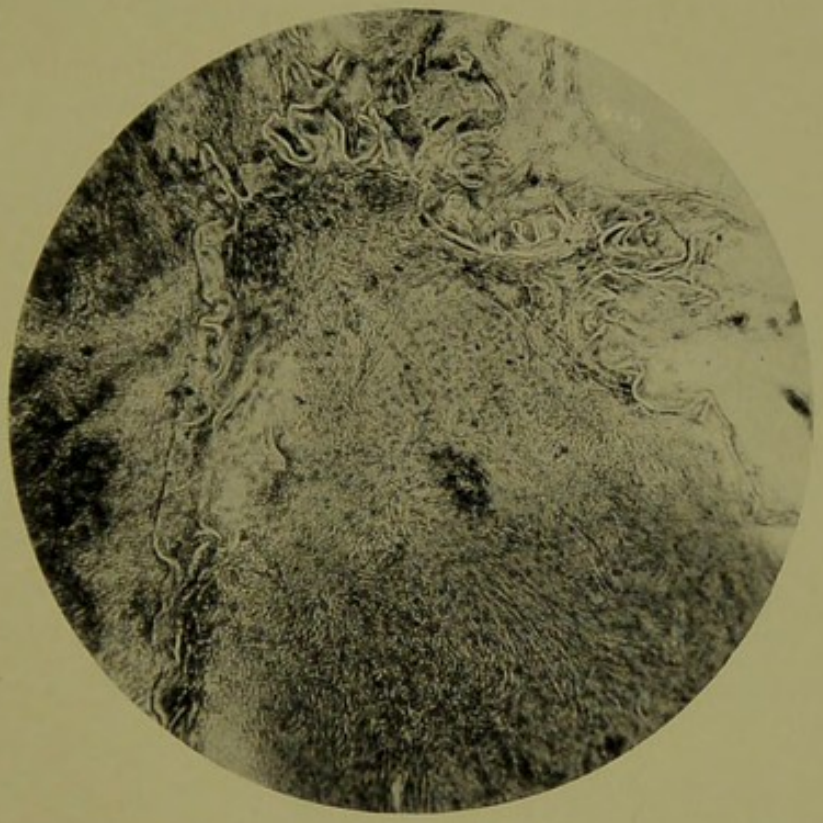


Fig. 3.

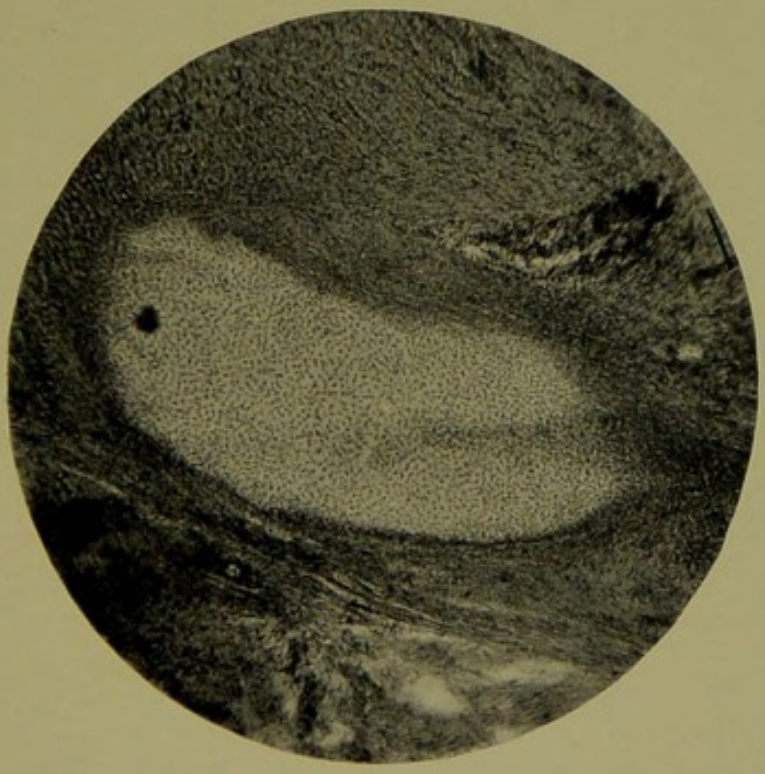


Fig. 4.

As already mentioned, no optic nerve can be distinguished in the specimen in the gap left in the sclerotic at its posterior part; some epithelial cells are seen to pass out at the margins.

Vitreous humour.—The main part of the gap is, however, filled by tissue composed of a loose fibrillar structure with cells with large nuclei scattered through it, but widely separated from one another and fairly evenly distributed. This tissue has numerous blood-vessels in it, surrounded by large lymphatic spaces. From the gap in the sclera at the posterior part this same tissue proceeds forwards, and forms the large, solid protuberance in the interior of the cyst (Pl. VI, fig. 1). It is probably atypically developed vitreous humour, in which the blood-vessels have remained persistent and continue to carry blood.

Lens.—The partition from the anterior wall joins this solid protuberance from behind. At the anterior part of the latter, where the two join, a much wrinkled lens capsule is to be seen (Pl. VII, fig. 3). Its anterior part is thrown into numerous convolutions, and has lining it epithelial cells in some places many layers thick. No well-developed lens-fibres are to be detected in the lens capsule. There is a considerable gap in the posterior part of the lens capsule, and through this gap blood-vessels from the atypical vitreous have evidently extended forwards into the lens-substance. The contents of the capsule has, therefore, become a mass of vascularised cellular tissue, in which it is very difficult to differentiate the epiblastic and mesoblastic elements. Some granules of calcareous material, staining deeply with the logwood, are seen scattered about in it.

Having endeavoured to describe as accurately as possible the appearances presented by this specimen in its various parts, it becomes necessary next to consider in what way the normal development of the eye has become arrested or changed to produce these strange

malformations. So complicated are they that the writer has found their interpretations beset with considerable difficulties. He has had the opportunity of examining pathologically several microphthalmic eyes having cystic protrusions connected with them, but none of these have been comparable with the present specimen.

In those previously examined there has always been a small, more or less spherical, globe surrounded by sclerotic, through a gap in which one or more folds of retina has protruded. It has been the distension of these folds which has constituted the cysts. In all of them the cyst has protruded from the lower part of the eyeball and extended forwards either into the lower part of the orbit or into the lower lid.

In the present case there is no definite structure surrounded by sclerotic which can be called a microphthalmic eyeball; the cyst is situated mainly in the upper eyelid and causes the whole conjunctival sac to be protruded through the palpebral aperture.

There are two cases recorded in which a congenital cyst, associated with so-called anophthalmos or microphthalmos, has been met with in the upper eyelid.

Mr. Simeon Snell, in the *Transactions* * of this Society for 1894, described the case of a girl, æt. six weeks, in whom a round, fluctuating, semi-transparent, bluish swelling, the size of a pigeon's egg, involved the whole of the right upper eyelid. It was dissected out, no eyeball was found, no histological examination was made.

Purtscher,† in the same year, described a cyst in the upper lid in a case of microphthalmos. He punctured the cyst and a yellow fluid escaped, it afterwards shrank. Later, another cyst formed at the bottom of the orbit. In this case, again, no histological examination was made.

These two cases, though resembling the one under consideration in having a cyst in the upper lid, did not present the peculiar extension of the whole conjunctival

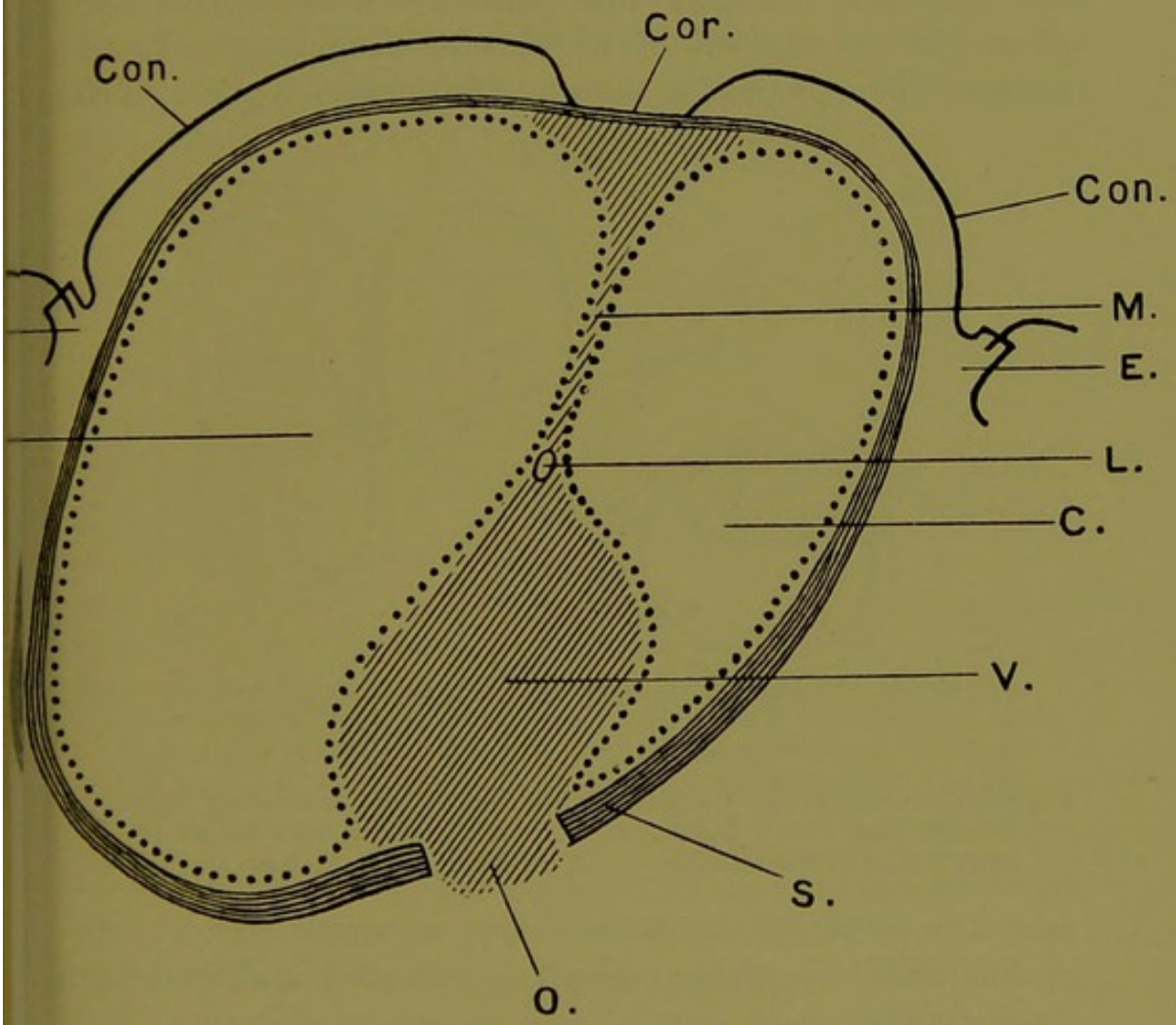
* Vol. XIV, p. 190.

† *Intern. klin. Rundschau*, No. 34, 1894.

sac through the palpebral aperture which formed in it such a striking feature.

The outer fibrous tissue wall of the cyst in this specimen would appear to be attenuated sclerotic; it is continuous with the tissue, which is undoubtedly sclerotic posteriorly, and it has the tendons of extra-ocular muscles inserted

Fig. 2.



Diagrammatic representation of a section through the cystic globe. Cor., cornea; Con., conjunctiva; E, eyelid; S, sclerotic; C, cavity of cyst; M, partition extending back from anterior wall of cyst; L, lens; V, atypically developed vitreous; O, gap in sclerotic corresponding to optic foramen.

into it. The circular piece anterior which comes to the surface, and which is devoid of any tissue corresponding to conjunctiva superficial and separate from it, is probably cornea.

It would appear, then, that the cyst is really an enormous distension of the whole globe.

The inner coat, corresponding to the retina, presents some most noteworthy features. That portion of the retina usually developed from the inner layer of the secondary optic vesicle nowhere lies in apposition with that usually developed from the outer layer. The anterior parts of the cyst are lined by the former—*i. e.* tissue composed of a delicate network of fibres with branching cells and nuclear bodies amongst them. The posterior parts of the

Fig. 3.

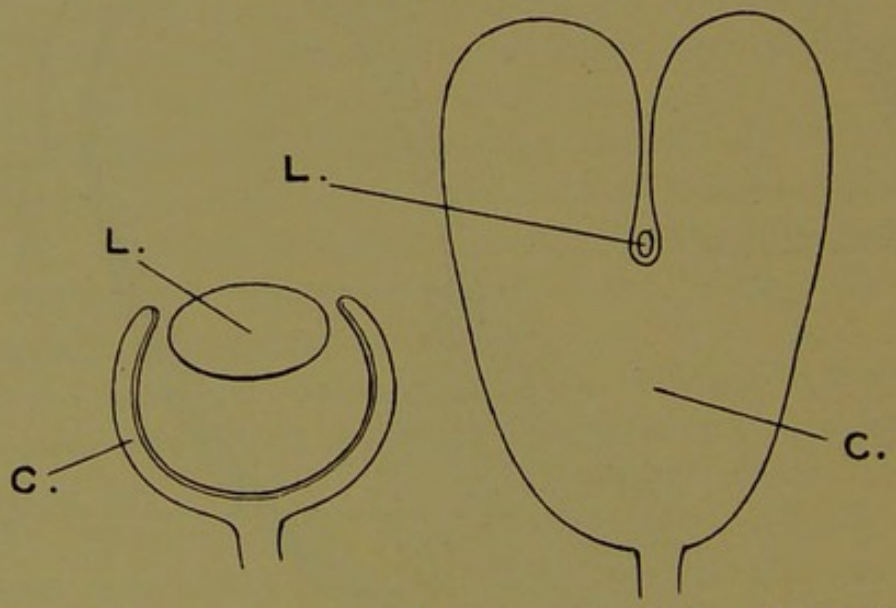


Diagram showing the normal involution of the secondary optic vesicle, and how a failure at its involution has resulted in the cyst found in the specimen. C, cavity of cyst; L, lens.

cyst are lined by the latter—*i. e.* a single row of epithelial cells in places deeply pigmented and in others unpigmented.

The cavity of the cyst is composed of a space between these two layers—that is, of the cavity of the primary optic vesicle.

The explanation of the formation of the cyst apparently is the imperfect involution of the primary optic vesicle to form the secondary optic vesicle.

There has been some attempt at involution both anteriorly and below.

Anteriorly we find the walls of the cyst prolonged backwards in the form of a fold, and at the bottom of the fold an attempt at the formation of a lens, so there must have been at one stage some downgrowth of surface epiblast.

Below we find the cavity of the cyst considerably encroached upon by a protruding mass of solid tissue, the histological characters of which suggest that it is mesoblastic tissue, which should have formed vitreous humour but which has developed in an atypical way. There being

FIG. 4.

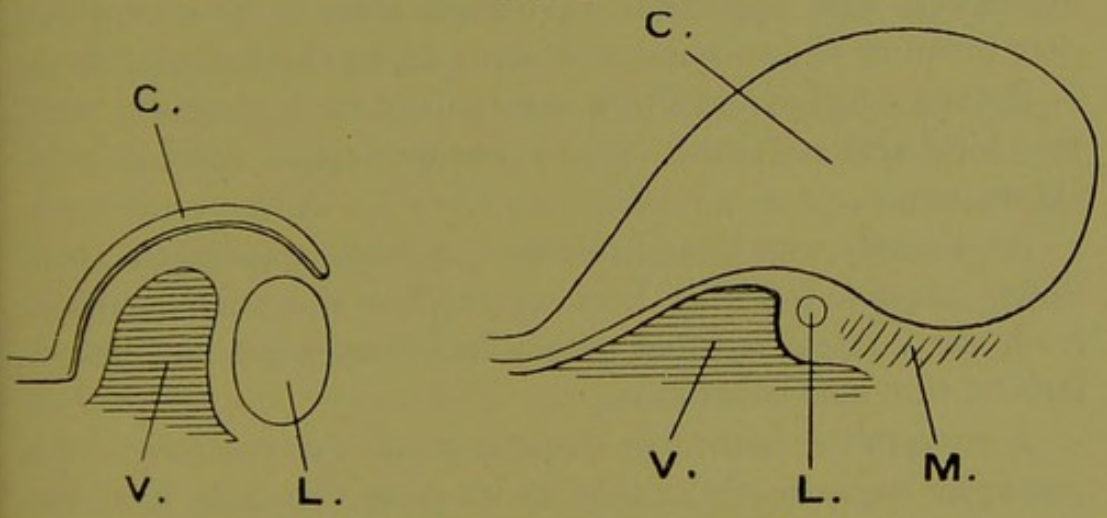


Diagram showing the normal upgrowth of mesoblastic tissue into the cavity of the secondary optic vesicle, and the effect of the upgrowth of this tissue on the cyst in this specimen. C, cavity of cyst; V, vitreous; L, lens; M, mesoblastic tissue anterior to lens.

no secondary optic vesicle for this to extend into, it has pushed forwards the posterior part of the inner wall of the cyst and is found lined by a single row of unpigmented cells everywhere except where it is in contact with the lens.

As no secondary optic vesicle formed, and the whole of that portion of the retina which normally forms its inner layer was lining the anterior part of the cyst, it is not surprising that no optic nerve developed.

Recognising the cyst to be due to a distension of the primary optic vesicle, we can understand why it mainly extended forwards and into the upper lid; below and behind

there was the upgrowing mesoblastic tissue which represented vitreous tending to check its enlargement in that direction.

The only specimen described, of which the writer knows, which at all resembles the condition found in this one was shown before this Society by Mr. Mayou* in 1904. In it there seemed to be a similar failure in the formation of the secondary optic vesicle. The specimen was removed from a child born at the seventh month who only lived twenty-four hours, and only very slight cystic distension had occurred. Had it lived, very probably this would have increased, and appearances, comparable to those in the case forming the subject of this paper, have been produced.

Before concluding there are points in connection with the lens and sclerotic which require some special consideration.

As already mentioned, there is a wide gap in the posterior capsule of the lens through which blood-vessels from the central hyaloid artery have extended and vascularised the lens-substance.

A congenital failure in development of a portion of the posterior capsule of the lens at its posterior pole has been met with in several other microphthalmic eyeballs.

The first case, so far as the writer knows, in which it was described was pictured and recorded by himself† in 1892. Mr. Parsons‡ met with the same condition in a specimen which he showed at this Society in 1902, and Mr. Mayou speaks of it in connection with lens in the specimen already referred to.

The writer has recently found another excellent example of it in a microphthalmic eye, with a persistent and patent hyaloid artery, of which he recorded a description several years ago, but in which at that time he overlooked the condition of the posterior capsule of the lens.

Behind the lens, where the central hyaloid artery

* *Ophth. Soc. Trans.*, vol. xxiv, p. 340.

† *Royal London Ophthalmic Hospital Rep.*, vol. xiii, p. 362.

‡ *Ophth. Soc. Trans.*, vol. xxii, p. 253.

divides up into branches, there is a mass of elongated cells and fibres, atypically developed vitreous. In sections through the centre of the lens the posterior capsule is seen to pass into this fibro-cellular tissue, to taper off and become lost. In sections passing a little to one side of the centre no break is met with in the continuity of the posterior capsule.

The vascularised fibro-cellular tissue at the back of the lens, evidently of mesoblastic origin, passes forwards through the gap in the lens capsule, as in the specimen forming the subject of this paper, invading the lens-substance. The amount of degeneration of the lens-fibres is not, however, quite so extensive.

In all the specimens presenting this congenital defect in the lens capsule, it seems to have been situated in the same position—*i. e.* at the posterior pole—and not to have extended very far either laterally or vertically.

There is considerable evidence to show that the lens capsule is the product of the epithelial cells lining it and is formed very early in fœtal life. The epithelial cells which line the posterior part of the lens vesicle are only present for a short time. They, soon after the formation of vesicle, lengthen out into lens-fibres.

The failure in the development of the lens capsule at the posterior pole is due probably to some failure in the secretion of it by these cells, the persistence of the central hyaloid artery, or its late disappearance, and the atypical development of the anterior portion of the vitreous, being most likely in some of the cases secondary to the defect in the capsule.

The condition, besides being of considerable embryological and pathological importance, presents features of clinical interest.

As is well known, in some cases of congenital cataract the lens appears to be shrunken, and when an attempt is made to tear through it with a needle it is found to be exceedingly tough. The shrunken condition and toughness may be easily accounted for by an invasion of, and

admixture with, the lens-substance of fibro-cellular tissue of mesoblastic origin through a gap in the capsule at the posterior surface.

The sclerotic, as already mentioned, for a short distance at the posterior part of this specimen presented much the normal characteristics. On one side, however, just before it began to thin out into the outer wall of the cyst, a small nodule of hyaline cartilage was found imbedded in it.

Hyaline cartilage has previously been met with in the sclerotic of congenitally malformed eyes.

The writer* showed this Society a specimen in 1897 of a congenitally microphthalmic eye with a cystic protrusion in which there was a large plate of hyaline cartilage imbedded in the sclera.

Mitvalski,† in the description of a somewhat similar microphthalmic eye with a cystic protrusion, says: "Where the scleral tissue passes over into the fibrous wall of the sac there is on each side in the entire length of the wall a mass of cartilage."

De Lapersonne‡ also mentions and figures a piece of hyaline cartilage imbedded in fibrous tissue lying between a microphthalmic eyeball and a cystic formation connected with it.

In many of the lower animals hyaline cartilage is met with in the sclerotic. In elasmobranch fish the sclerotic consists of hyaline cartilage from the sclero-corneal margin right back to the optic nerve with only a small amount of fibrous tissue on its outer surface. In most teleostean fish there is hyaline cartilage anteriorly extending up to the margin of the cornea, but posteriorly there is only fibrous tissue, or fibrous tissue with scattered patches of cartilage in it. In birds and some reptiles there is a cup of hyaline cartilage posteriorly, extending forwards beyond the level of the ora serrata, and anteriorly to it are plates of bone.

* *Ophth. Soc. Trans.*, vol. xvii, p. 264.

† *Arch. of Ophth.*, vol. xxii, p. 355.

‡ *Arch. d'Ophtalmol.*, vol. xi, p. 207.

In some reptiles the sclerotic is fibrous throughout. In all mammals, with the exception of the ornithorrhynchus and echidna, the sclerotic is entirely fibrous. In the two exceptional mammals it contains a cup-shaped piece of hyaline cartilage at the posterior part extending forwards as far as the ora serrata, and having fibrous tissue on its inner and outer surfaces.

If, then, we are to regard the formation of hyaline cartilage in these congenitally malformed human eyes as an example of the reversion of a tissue to a more primitive or ancestral type, we should have to go right back to the ornithorrhynchus or echidna before we find it existing as the normal condition. (*May 3rd*, 1906.)

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