Microphthalmos with cystic protrusion from the globe / by E. Treacher Collins.

Reprinted from Vol. XVII of the Ophthalmological Society's 'Transactions.'

Microphthalmos with cystic protrusion from the globe.

By E. Treacher Collins.

In 1893 I described to this Society three congenitally microphthalmic eyes I had examined microscopically, in which there was a protrusion of retinal tissue through a gap in the sclerotic. I had previously described another case of a similar character recorded by Mr. Lang in the 'Royal London Ophthalmic Hospital Reports' (vol. xii, p. 289). Since 1893 I have examined three more microphthalmic eyes presenting this abnormality,—one removed by Mr. J. R. Rolston, who kindly entrusted me with the pathological examination of the specimen, and two I have removed myself from patients under my care at the Moorfields Hospital, which I have to show you this evening.

Having thus had considerable material upon which to study this rare form of congenital malformation, I may presume to generalise, and propose to give you a description of it drawn chiefly from the seven eyes I have examined, but also partly from those described by others.

Though the patients presenting this abnormality of the eye are frequently the subjects of malformations of other parts of the body (Kundrat's cases [1]; Case 1, 'Trans. Ophth. Soc.,' vol. xiii; Case 2 in this paper), it also occurs in individuals who are in all other respects apparently perfectly healthy and well formed (Case 2, 'Trans. Ophth. Soc.,' vol. xiii; Case 1 in this paper).

Though all my specimens are alike in one particular, namely, in being microphthalmic eyes with a protrusion

of retinal tissue through a gap in the choroid and sclerotic, the differences which they present, both as regards the character of the protrusion, and in the state of development or mal-development of the other structures of the eyeball, are truly remarkable.

The protuberant portion of retina consists of varying amounts, from simply a knuckle of tissue (Case 1, 'Trans. Ophth. Soc.,' vol. xiii) to quite two thirds of that membrane (Case 1 in this paper, Fig. 1). In some cases it forms solid mass surrounded and intermixed with bands of fibrous tissue (Case 1, 'Trans. Ophth. Soc.,' vol. xiii; Case 1 in this paper, Fig. 1). In others, folds of retina have become distended into one or more fluid-containing cystic protrusions, which are bounded externally by a fibrous-tissue coat, and have a very lobulated outline (Lang's [2] case; Case 2, 'Trans. Ophth. Soc.,' vol. xiii; Rolston's case; Case 2 in this paper).

The degree of development of the protuberant retina is also very different in different cases. When folds of it have become distended in the form of cystic protrusions, it may be so atrophied as merely to be represented by a few groups of nuclear bodies and a little retiform tissue (Lang's [2] case; Case 2, 'Trans. Ophth. Soc.,' vol. xiii). On the other hand, it may remain so well developed that some of its different layers are easily distinguished (Case 2 in this paper). In these latter cases it is found that sometimes the inner surface of the retina is turned towards the interior of the cyst (Rindfleisch [3]) and at other times just the reverse, the inner surface of the retina being directed towards the fibrous outer wall of the cyst (Case 2 in this paper; De Lapersonne [4]; Czermak [5]).

The region of the globe in which the protrusion is situated is nearly always its lower and posterior part, sometimes a little further forwards than others. In one case, besides the gap in the sclerotic there was a considerable deficiency in the outer sheath of the optic nerve (Rolston's case).

Speaking generally, the larger the amount of the retinal protrusion, the smaller and more imperfectly developed is the eyeball. When the protrusion is not very extensive the eyeball may present little external evidence of malformation beyond its defect in size. The lens and vitreous may also appear quite normal (Case 1, 'Trans. Ophth. Soc.,' vol. xiii, R. eye). On the other hand, there are numerous congenital defects of the eye which may exist together with this protrusion of retina.

The cornea may be exceedingly small, out of proportion to the rest of the globe, and in addition to being small, opaque, with blood-vessels coursing through its anterior layers (Lang's [2] case; Case 2, 'Trans. Ophth. Soc.,' vol. xiii; Cases 1 and 2 in this paper). The fœtal cornea in no stage of its development has any blood-vessels formed in it, so that this vascularisation of the cornea is not simply a persistence of any fœtal condition. In one case the cornea was abnormally large for the size of the globe, and globular, as in buphthalmos (Rolston's case).

The sclerotic is usually unduly thick, and in some cases there exist embedded in it plates or nodules of well-developed hyaline cartilage (Case 2 in this paper (Fig. 2); De Lapersonne [4]; Mitvalsky [6]). This formation of hyaline cartilage in the sclerotic is very remarkable, and the only thing I know with which to associate it is the sclerotic of a bird in which normally hyaline cartilage exists, somewhat comparable in its arrangement to that in one of my cases.*

The iris, besides being small, may present no congenital deficiency (Case 1 in this paper; Case 1, 'Trans. Ophth. Soc.,' vol. xiii, R. eye). Bands of pupillary membrane may be persistent (Case 1 in this paper; Case 1, 'Trans. Ophth. Soc.,' vol. xiii, R. eye), or there

* Mr. Marcus Gunn has called my attention to a description of the eye of the *Ornithorhynchus paradoxus*, or duck-mole, published by him in the 'Journal of Anatomy and Physiology,' vol. xviii, p. 400, in which he mentions plates of hyaline cartilage existing in the sclerotic.

may be a coloboma present (Lang's case; Case 1, 'Trans. Ophth. Soc.,' vol. xiii, L. eye; Case 2, vol. xiii; Rolston's case).

The ciliary processes often become drawn backwards, being elongated and thin (Case 1, 'Trans. Ophth. Soc.,' vol. xiii, L. eye; Rolston's case; Case 2 in this paper). The pigment epithelium overlying them may show disturbance (Case 1 in this paper; Rolston's case), and sometimes the unpigmented layer of the *pars ciliaris retinæ* is raised in folds, which shows imperfect coaptation of the two layers of the secondary optic vesicle in that position (Case 2 in this paper).

The lens is generally imperfectly developed and cataractous, usually spherical in shape (Lang's case [2]; Case 1, 'Trans. Ophth. Soc.,' vol. xiii, L. eye; Case 2, vol. xiii; Rolston's case; Case 1 in this paper), and sometimes displaced.

The vitreous humour is often imperfectly formed, being in part replaced by a kind of fibrous tissue which may have blood-vessels coursing through it (Case 1, 'Trans. Ophth. Soc.,' vol. xiii, L. eye; Rolston's case; Case 2 in this paper). This formation of fibrous tissue in place of vitreous humour, apart from any signs of inflammation, is a condition to which I particularly wish to direct your attention. It has been termed by Hess (7) "atypical development of the vitreous humour,"—that is to say, the mesoblast which should have formed vitreous has developed in an atypical way and formed fibrous tissue. I have met with this malformed vitreous humour, in eyes which I have examined pathologically, in various positions. I have found it forming a membrane of varying thickness behind the lens, into which sometimes the central hyaloid artery has passed and divided up; as irregular bands passing forwards from the optic disc; as a thick mass on the inner surface of the globe in the region of the ocular cleft; and in one of the specimens which I am bringing before you for the first time this evening, it would seem, that the mesoblast which should

have formed the vitreous had almost entirely failed to pass into the globe, but had, instead, developed into fibrous tissue beneath it. With this fibrous tissue the protuberant and folded retina has become intimately intermixed.

As I have said, I particularly wish to draw your attention to the possibility of this atypical development of the vitreous humour, because it helps to explain a group of cases in which the ophthalmoscopic appearances are often rather perplexing. Scattered throughout ophthalmic literature will be found the records of a number of eyes, the sight of which, the history states, has been defective since birth; which, when examined ophthalmoscopically, are found to have irregular bands of fibrous tissue in the vitreous chamber, often with blood-vessels coursing through them.

Some have thought these cases to be of the nature of what is termed "retinitis proliferans," to the ophthalmoscopical characters of which they frequently bear some resemblance; from which, however, they differ in not following on or being associated with any intra-ocular hæmorrhage, in being non-progressive, and in dating from birth. Others have attributed these bands of fibrous tissue in the vitreous chamber to persistence of the central artery of the vitreous or the canal of Cloquet; but anyone who has seen under the microscope what the central hyaloid artery and the canal of Cloquet consist of in the fœtal eye, will at once realise that the mere persistence of these structures will not account for the dense bands and collections of fibrous tissue which are sometimes seen.

Returning now from this digression concerning atypical development of the vitreous humour to the condition of the microphthalmic eyes with retinal protrusions, I find the retina in these eyes, except in the region of the protrusion, may be situated in its normal position. More frequently it is raised in rucks from the pigment epithelial layer beneath, which rucks often become much folded. On

the inner surface of the choroid are frequently seen nodules of hyaline substance lying amongst the pigment epithelial cells (Case 2, 'Trans. Ophth. Soc.,' vol. xiii), and also sometimes a formation of bone (Rolston's case). Hyaline nodules of a similar character are also sometimes met with in the retinal tissue in the interior of the cystic protrusions where no choroid is present (Case 2, 'Trans. Ophth. Soc.,' vol. xiii; Rolston's case; Hess [7]).

In my former communication on this subject to this Society, I referred to the various theories which have been put forward to explain these protrusions of retinal tissue through gaps in the other tunics of the globe. I need not repeat them here, but content myself with the following statement. In all the specimens which I have examined the changes are best explained by considering the inner layer of the secondary optic vesicle to have become unusually rucked and folded, due to some imperfect development of the vitreous humour or delayed closure of the cleft; that, as the result of this rucking and folding, a portion of the inner layer of the secondary optic vesicle has become extruded through the fœtal ocular cleft into the subjacent mesoblast, where in some cases it has subsequently become expanded into one or more fluid-containing cysts.

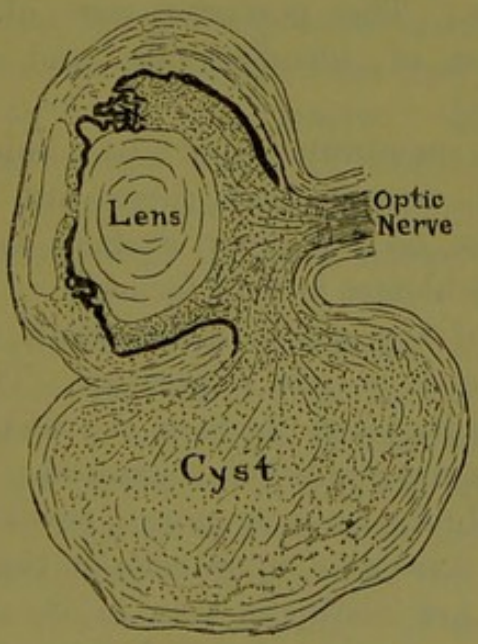
Case 1 (Clinical notes by Mr. Fischer, House Surgeon).—William M—, æt. 28, admitted to Moorfields Hospital August 10th, 1896. Patient stated that his right eye had been small and shrunken since birth. A doctor who examined it on the third day after birth said it was blind. The condition was attributed to a fright his mother received while pregnant. Recently it has been very painful, the pain coming on in recurrent attacks, and at times the sight of his left eye getting misty.

The right globe was small, and almost completely hidden by overlapping conjunctiva. The left eye was perfectly healthy, and V. $= \frac{6}{6}$. The patient presented no other congenital abnormality.

On the day of admission the right eyeball with a cyst attached to the back of it was excised.

Pathological examination.—Macroscopical appearances.—The eyeball measured only 8 mm. antero-posteriorly and 8 mm. laterally. In front was a small clear cornea, measuring 3 mm. laterally; and behind, the point of section of the optic nerve could be recognised. Attached by a broad pedicle to the lower and posterior part of the globe was a protuberance, the wall of which was continuous with the sclerotic (Fig. 1). In front a deep sulcus separated this mass from the globe. Posteriorly the protuberant mass had been cut into in its removal, and it was seen to be a cyst filled with greyish lobulated contents. The cyst measured 17 mm. antero-posteriorly, and 8 mm. laterally. On section of the globe the cornea, though small, was seen to be of about the normal

Fig. 1.



thickness. The anterior chamber was well formed but shallow. The iris and ciliary body, though small, were not absent in any part of their circumference.

The interior of the globe behind the iris was almost entirely filled by the lens, which was spherical in shape,

measuring 2 mm. laterally and antero-posteriorly. It had some calcareous deposit in it. No break was observed in the continuity of the sclerotic from the margin of the cornea to the optic nerve at the upper part of the globe. In the lower part a little behind the equator the sclerotic appeared to end abruptly, and there was a gap in it from this point to the lower border of the optic nerve (Fig. 1). Where the sclerotic was deficient, the cystic mass above referred to was attached to the globe, its outer wall being continuous with sclerotic. There was hardly any retina in the globe ; nearly all of it was contained in the mass below it, where it formed the folded membranous contents of the cyst.

Microscopical appearances.—The epithelium on the anterior surface of the cornea is thick, and its base line very irregular. There is no anterior elastic lamina. In the anterior part of the substantia propria there are some variously sized, thin-walled blood-vessels, and an excess of cells. The posterior part of the substantia propria is free of blood-vessels, and more regularly laminated.

Descemet's membrane and its lining endothelium appear normal, except that there are numerous little hyaline nodules on its inner surface at the periphery. The canal of Schlemm is well formed.

The tissue of the iris is denser than normal ; a rudimentary sphincter muscle is present. The two layers of pigment on its posterior surface are distinctly shown, as they have become somewhat separated. Portions of the pupillary membrane have remained persistent.

The ciliary muscle is more cellular than usual ; longitudinal fibres are distinctly seen in it. In its posterior part there is some round-celled tissue. The pigment epithelium on the inner surface of the non-plicated portion of the ciliary body has undergone proliferation, and many tubular processes of pigmented cells are seen cut, some longitudinally, and some transversely. On the inner surface of the pigment epithelium of the ciliary body,

instead of a single row of cells there is a sort of degenerated retinal tissue with calcareous granules in it. This tissue is continuous with the retina behind the lens. The choroid at the upper part is thick and vascular. Below it cannot be traced very far before the break occurs in the continuity of it, and of the sclerotic. Round the edge of the termination of the sclerotic the choroid and pigment epithelium turn and extend for a short distance.

The sclerotic at the lower part of the globe is continuous with a mass of fibrous tissue, which forms the anterior part of the thick outer wall of the cyst; its limits cannot be defined. The retina does not present its normal layers, but consists of branching cells, bundles of fibres like Müller's fibres, and collections of nuclear bodies. The gap in the sclerotic and choroid is filled with this altered retinal tissue, and continuous with it are the contents of the cyst, which bear only a slight resemblance to the retina, being composed of a dense network of fibres and nuclear bodies, with here and there patches of deeply staining calcareous matter. Mixed up with this tissue are numerous bands of fibrous tissue. The posterior wall of the cyst is not shown in the sections, nor is the point of entrance of the optic nerve.

Case 2.—William L—, æt. 14 weeks, was brought to me at the Moorfields Hospital by his mother on March 8th, 1897, on account of a congenital defect of his left eye. She stated that the condition of his left eye when born was exactly as it is now, except that there was a red growth at the outer angle of the lids the size of a pea, which the doctor who was attending her removed with a piece of silk. No instruments were used at the birth. There had not at any time been more than a slight discharge from the eye. The lids on the left side did not meet at the outer canthus, there was a small gap left between them, where injected conjunctival tissue was exposed.

On separation of the lids an exceedingly small eyeball was seen ; what appeared to be the cornea, from the position of the insertion of the conjunctiva, was a very small opaque patch. There was a mass of firm tissue felt adherent to the lower and outer part of the rudimentary globe.

The child's right eye was normal. He had a large hairy mole on the left temple, and an oval pigmented patch of skin over the left parietal bone.

On March 15th, 1897, I removed the small eyeball with the mass attached to it, because it was absolutely useless, and would have prevented his wearing a glass eye. I afterwards pared the edges of the lids at the outer canthus, and united them over the exposed conjunctiva.

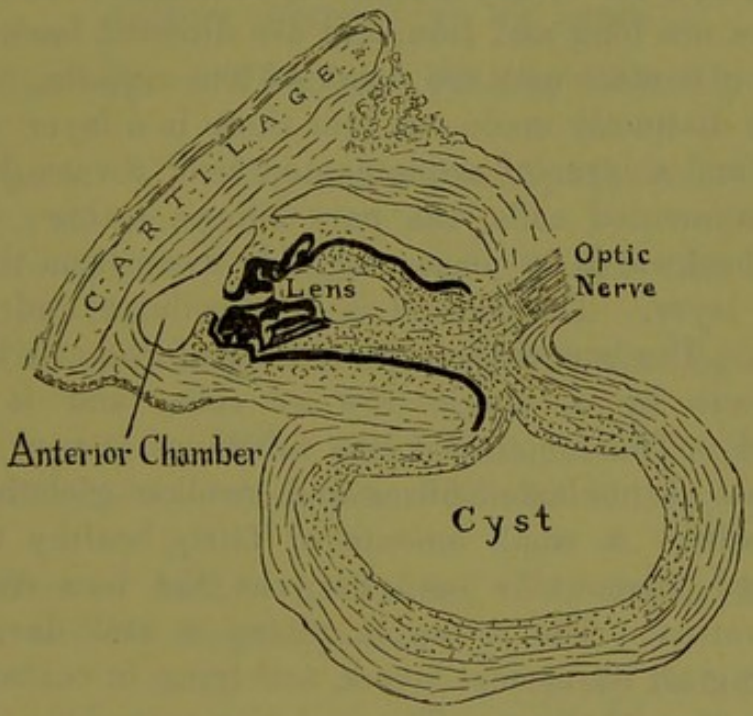
Pathological examination : microscopical and macroscopical appearances.—The small spot on the surface of the globe, around which the fringe of conjunctiva was attached, and which appeared to be the rudimentary opaque cornea, measured 4 mm. by 1 mm. Above and behind it, in the wall of the globe, a hard flat plate of tissue was felt, behind which again the optic nerve was seen where it had been cut across. Attached to the lower part of the globe in front of the optic nerve was a lobulated thin-walled cyst, the front wall of which had been cut into, and from which, on pressure, a clear straw-coloured fluid was made to exude.

After the specimen had been hardened in formol it was opened by an antero-posterior section, and subsequently microscopic sections were cut.

The area on the surface of the globe, around which the conjunctiva was attached, and where the anterior epithelium of the cornea is situated, does not extend over nearly the whole of the substantia propria, but only one side of it. This surface epithelium is prolonged into the subjacent fibrous tissue in the form of finger-like processes, so that it presents a very irregular base line ; beneath it there is no sign of any anterior elastic lamina.

In front of a large part of the cornea, and partly embedded in it and partly in the sclerotic, is a broad plate of hyaline cartilage. This cartilage ends anteriorly in a rounded extremity amongst the fibrous tissue beneath the epithelium of the conjunctiva. Posteriorly it also ends in a rounded extremity surrounded by fibrous tissue, and projects as a sort of spur from the sclerotic about the

Fig. 2.



equator of globe, there being fatty tissue in the recess left between the spur and the sclerotic (Fig. 2). There is a second piece of hyaline cartilage in the sclerotic on the opposite side of the globe to the other, which commences posteriorly in a rounded extremity a short distance from the optic nerve; the anterior termination of it is not shown in the sections. In some sections this last piece of cartilage is seen as a circular patch lying in the fibrous tissue between the eyeball and the cyst. The substantia propria of the cornea is fairly regularly laminated; amongst its layers immediately beneath the plate of hyaline cartilage are some blood-vessels. Near the sclero-corneal margin on the side furthest from the surface

epithelium there is a cleft in the fibrous tissue, containing adipose tissue. The posterior elastic lamina and its lining endothelium are well formed; it is much wrinkled.

There is a shallow anterior chamber. The iris, though very small, is fairly well developed; there are blood-vessels in its stroma, and a rudimentary sphincter muscle; the pigmented epithelium on its posterior surface is folded and irregular. The ciliary body, for the size of the globe, is large, and the ciliary muscle very cellular. The ciliary processes are long and thin, and are directed backwards; they lie in contact with the wrinkled lens capsule. It can be quite distinctly made out that there is a layer of pigmented and a layer of unpigmented cells covering them; the unpigmented cells, the pars ciliaris retinæ, is prolonged backwards in places in folds away from the pigmented layer. The choroid is very cellular and unpigmented. The lens capsule is well developed; it is lined throughout by a single row of cells, and is much wrinkled. The amount of lens substance is very small; it consists of nucleated fibres and swollen globular epithelial cells. A small amount of fairly healthy though condensed vitreous is seen between the lens and the folded retina. The retina in places is well developed, presenting all its normal layers, and lying in contact with its pigment epithelium. The posterior part of the globe is filled with well-developed, much-folded retina. A section passing across the neck of the cyst shows retinal tissue passing out from the interior of the globe, through a narrow gap in the sclerotic at the margin of the optic nerve (Fig. 2).

There is some pigment in the retinal tissue passing through the gap, which does not, however, extend further than the neck of the cyst.

The cyst walls are very irregular, and present many projections and recesses. The cyst everywhere has two coats, an outer of fibrous tissue continuous with the sclerotic, and an inner of ill-developed retinal tissue. In this inner coat the layers are sufficiently differentiated to

make it evident that the inner surface of the retina is directed outwards, and the outer surface towards the interior of the cyst. Where it is best developed the nerve-fibre layer and the ganglion-cell layer can be distinguished; the granular layers appear to have run together: the membrana limitans externa is present, but there are no rods or cones.

Authors referred to in the paper.

(1) *Kundrat.* 'Wien. med. Blätter,' Nos. 51, 52, and 53.

(2) *Lang, W.* 'Royal London Ophthalmic Hospital Reports,' vol. xii, p. 287.

(3) *Rindfleisch.* 'Archiv für Ophth.,' Bd. xxxvii, Abth. 3, s. 192.

(4) *De Lapersonne.* 'Archives d'Ophtalmologie,' t. xi, p. 207.

(5) *Czermak.* 'Wien. klin. Woch.,' 1891, p. 448.

(6) *Mitvalsky.* 'Archives of Ophthalmology,' vol. xxii, p. 355.

(7) *Hess.* 'Archiv f. Ophth.,' Bd. xxxvi, Abth. 1, s. 135; and Bd. xxxviii, Abth. 3, s. 93.

(*July 2nd*, 1897.)

PRINTED BY ADLARD AND SON,
BARTHOLOMEW CLOSE, E.C., AND 20 HANOVER SQUARE, W.