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A RARE CONGENITAL ABNORMALITY OF THE EYE

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THE following case is reported, since, although it belongs to a group of congenital defects (fibro-lipoma, dermo-lipoma or dermoid of the cornea) which are relatively common and well known, it apparently furnishes an example of a very rare variety of the condition. As far as can be ascertained only three cases at all similar to the present have been described.

According to Stargardt¹ congenital abnormalities of the cornea of a fibro-lipomatous nature can be divided into four groups, which depend embryologically for their explanation on the period of development at which they arose. It appears from a survey of the literature that the occurrence of such anomalies is in inverse ratio to their earliness of origin. This is true of most congenital defects and is more or less self-evident, since the earlier in ontogeny that developmental aberration becomes evident the less good is the prospect of survival of the embryo.

All types of so-called congenital dermoid of the cornea can be referred to abnormal development of the mesoblast lying between the anterior rim of the optic cup and the surface ectoderm.

The first and rarest malformation can be referred to the period of growth immediately before the formation of the lens (in an embryo of about 4.5 mm.). Its definitive condition is characterised by the substitution for the cornea of a mass of fibrous tissue usually containing fat. The eye is always

microphthalmic and the membrane of Descemet, the anterior chamber, the iris and the lens are absent. As far as I am aware only three undoubted cases of this anomaly have been recorded. The present case affords a fourth example of this condition.

The cases in the second group arise during the process of formation of the lens, probably before its hyaline capsule is complete (*circa* 12 mm.). They show also the substitution of a mass of fibro-lipomatous tissue for the cornea, but this is associated with a poorly developed or even rudimentary lens. Traces of iris may be recognised in these cases.

In the third group the defect arises after the formation of the lens and anterior chamber and consists merely of the substitution of the abnormal mass for the whole or part of the cornea.

The fourth group contains the well-known limbal dermoids which are relatively common and arise as purely local anomalies of differentiation of a portion of the already forming cornea and sclera.

The present specimen will now be described and comparison made with the other recorded similar conditions.

I am indebted to Colonel J. N. Duggan of the Sir C. J. Ophthalmic Hospital, Bombay, for permission to publish the case. He has also kindly furnished me with the clinical history and the photographs of the child (Fig. 1.)

The child, a female Hindu of four months old, was brought to the hospital on January 25, 1927, on account of a congenital tumour of the left eyeball. The appearance of the child is shown in Fig. 1. In place of the left eyeball was a slightly movable pedunculated rounded swelling measuring about two inches in diameter. Posteriorly it was attached to a fibrous pedicle of about $\frac{3}{4}$ inch in diameter. The eyelids and eyelashes were normal. The former were found nearly encircling the stalk just behind the posterior surface of the tumour. The whole mass moved to some extent with the movements of the other eye, which was normal. On palpation the tumour felt hard. The surface covering resembled skin which passed imperceptibly into the conjunctiva covering the orbital cavity and the stalk. There was no sign or history of inflammation. The mass was removed under general anaesthesia. It could not be separated from the eyeball, of which it formed the major part. The stalk was ligated and cut. No abnormality in the orbital cavity was found. The child made an uninterrupted recovery.

The tumour was fixed in formol saline. It was divided in the sagittal plane. The pedicle, presumably the optic nerve, was attached to its posterior pole. The portion containing this was retained in the Pathology Department at Bombay. The other half

FIG. 1.

Appearance of the patient. (From a photograph by Colonel Duggan).

was brought to England by Colonel Duggan and sectioned in the laboratory at Moorfields.

The general topography of the specimen is shown in Fig. 2 which is three times the size after fixation. The actual specimen measured after fixation, 31.5 mm. $\times$ 25 mm. in its two greatest diameters.

The anterior portion of the mass (at A) is covered with skin. The mass itself is composed of fibrous tissue, containing areas of fat and in some situations showing myxomatous degeneration. This is most marked in the pale areas at M, which appear marked off by their less dense staining from the rest of the tumour. The posterior third of the tumour is occupied by a cavity, V, which represents the vitreous chamber. At the posterior pole, P, sclerotic

and choroid, normal in structure but rather thick, can be recognised. There is no trace of conjunctiva. The whole mass as far back as S is covered with pigmented skin. There is no trace of lens, anterior chamber, iris stroma or ciliary muscle. The neuro-epithelial elements derived from the primary optic vesicle are easily

FIG. 2.

Gross morphology of the specimen. The rectangles 3, 4, 5, 6, 7, and 8 mark the situation of the sections shown in the subsequent figures.

recognisable, though they show aberrant development. It is much to be regretted that I was not able to obtain the pedicle for section. In the posterior part of the vitreous chamber there is a mass of blood clot, presumably operative and sub-retinal, and most of the retina has become detached and lost. Traces of it, showing an abnormally rich vascularisation, could be found in some of the sections.

The tumour is therefore obviously an aberrance of differentiation of the mesoblast lying between the optic cup and the surface ectoderm. It must have begun to arise very early, since it has inhibited the development of the lens entirely. The other accompanying abnormalities (skin instead of conjunctiva, faulty retinal differentiation and absence of iris) are secondary to this primary fault and can be explained as attempted adaptations to altered environment. Whether the mesoderm of the foetal intra-ocular vessels was also faulty cannot be determined from the portion of the specimen available, but it has been shown to be abnormally persistent in other cases of the same nature and it is possible that it was so in this case also, as there is evidence of excessive intra-ocular vascularity. No definite vitreous vessels were, however, recognised in either half.

Certain regions of the specimen are of special interest, some as showing the presence of abnormal tissues, others as illustrating altered growth in response to altered environment. (The small rectangles in Fig. 2 indicate the areas shown under higher magnification in subsequent figures.)

FIG. 3.

Fibro-lipomatous portion of tumour (at 3, in Fig. 2) showing cellular fat.

Immediately beneath the skin covering the surface of the tumour is a layer of fibrous tissue containing blood vessels and sweat glands. No hairs were seen. Deep to this again is a large area of fibro-fatty tissue similar to that found in any fibro-lipoma. At M the fibrous tissue predominates over the fat and shows myxomatous degeneration. Posterior to M there is still more fibrous tissue as compared with fat, and what fat there is is peculiarly cellular. Fig. 3 shows a portion of this cellular fat. It is lying in irregular spaces between bundles of rather primitive fibrous tissue containing blood vessels. The fatty tissue bears a very strong resemblance to bone marrow and when its cells are examined under a high power, a great diversity of form becomes apparent and it is possible to recognise with a fair amount of certainty the presence of myelocytes, megakaryocytes, basophile and eosinophile leucocytes, lymphocytes, normoblasts and free red cells. This myeloid reaction of the fat, though interesting, cannot be used, as might at first sight seem possible, to support any theory of teratomatous or blastomatous origin of the tumour, since it must be remembered that isolated islands of marrow-like tissue are of common occurrence in infants. They are found in the viscera, liver, spleen and kidney and in other situations in normal children and are merely an expression of the great haematopoietic capacity of young children. There is no *a priori* reason why they should not occur in a patch of atypically differentiated tissue as easily as in normal tissue.

The vagaries of the neural epiblast in relation to the posterior surface of the fibro-lipoma are worthy of study as they illustrate so well the pluripotentiality of the cells of the optic vesicle in early stages of development.

If the lining of the vitreous cavity be examined at 4 (in Fig. 2) it will be seen that in the situation where one might expect to find traces of ciliary processes there is instead a transition, in the outer layer of the optic cup, to a primitive type of retina. The fate of the inner layer cannot be traced, but the outer layer (*i.e.*, that which should have formed the anterior or outer layer of the epithelium) shows an interesting series of changes as one passes along the posterior surface of the tumour. Fig. 4 shows it forming a neuro-epithelium (like that found at the posterior pole in a 10 mm. human embryo) in which a marginal and a mantle layer can be recognised. 1 marks the nuclei of the mantle layer, 2 the marginal layer, 4 the point of transition from this to 5, the normal pigment epithelium. 3 is the fibrous tissue of the tumour. It is to be noted that, since the differentiation is taking place in the *outer* wall of the original optic cup the marginal layer is turned *away* from the vitreous cavity. As one proceeds along the posterior surface of the tumour one encounters a more advanced stage of differentiation. At 5 (Fig. 2) the tissue shows an inner and outer neuroblastic

layer such as is found normally at the posterior pole at 17-20 mm. Fig. 5 represents this. 1 is the "outer" neuroblastic layer, 2 the fibre layer of Chievitz, 3 the "inner" neuroblastic layer (in this abnormal situation "inner" and "outer" are of course reversed) and 4 an invasion by blood vessels from the fibrous mass. Between 4 and 6 (Fig. 2) there are large areas of very

FIG. 4.

Abnormally differentiated retina. For reference numbers see text.

FIG. 5.

Abnormal retina showing inner and outer neuroblastic layers. For reference numbers see text.

FIG. 6.

Abnormal retina showing three cell layers. See text.

atypical arrangement, but at 6 there is a small fold of tissue on which still further differentiation has occurred. Fig. 6 shows this. The outermost layer 1 is extremely fibrillar and represents the nerve fibre layer. 2 shows an irregular layer of isolated cells, some of which distinctly resemble ganglion cells. Deep to them at 3 is a layer of deeply staining nuclei (inner nuclear layer) separated by a narrow interval (4) from 5, a layer of large nuclei with open mesh and prominent centrosomes. These are oval, primitive in shape and are associated with curious large protoplasmic or cuticular protuberances towards and through a thin structureless membrane. The whole is reminiscent of the retina at 50-65 mm.; the rod and cone-like protuberances are larger than they would ever normally be, but their relation to the thin membrane (? external limiting membrane) is suggestive that they are morphologically comparable structures. They point, of course, towards the cavity of the primary optic vesicle.

Fig. 7 shows another atypical patch of tissue in which a somewhat primitive neuro-epithelium has undergone cystic degeneration. The patch of tissue is very reminiscent of the small retinal cysts often found at the ora serrata as a senile change in slightly degenerate eyes.

So far all the structures apparent in the neuro-epithelial tissue of the anterior part of the eye have been abnormal. It remains to

FIG. 7.

Cystic degeneration.

FIG. 8.

Neural tissue invading fibrous tissue. 1 points to neural epiblast, 2 to fibrous tissue.

ascertain whether any trace of the neural part of the iris, namely pigment epithelium or unstriped ectodermal muscle fibres can be discovered. Pigment is certainly present, but it occurs in isolated cells irregularly scattered among the abortive retinal elements and not anywhere as a definite layer. The question of unstriped muscle is more complicated. Sections stained with van Gieson's stain show (at 8 in Fig. 2 and for a short distance on either side) very clearly the distinction between the ectodermal and mesodermal elements in this region. The frankly retinal portion is easy to understand, but between it and the definite fibrous tissue is an irregular zone of yellow stained tissue markedly resembling unstriped muscle but directly continuous with the retinal portion. This tissue is invading the fibrous tissue and breaking it up into bundles. In places it appears syncytial, in others elongated cells with rod-shaped nuclei can be made out. It seems possible that

it represents an abortive attempt on the part of the neural ectoderm to form a sphincter. Fig. 8 shows it spreading into the fibrous tissue. 1 points to the neural tissue, 2 to the fibrous.

This case belongs to that group of congenital fibro-lipomata of the cornea which arise by an aberration of mesoderm at or about the fourth week of development. They are, according to Stargardt, true congenital anomalies (*Missbildung*) and should not properly be classed as new growths (*Geschwulst*). From a pathological point of view they belong to the group of monophyllic teratoblastomata as they arise primarily from the aberration of one germ layer only. The skin and abortive retina are merely changes in surrounding structures secondary to the primary anomaly. Such cases appear to be of extreme rarity. The three hitherto recorded which were undoubtedly of the same nature are those of Swanzy and Leber² (1871), Hanke³ (1904), and Stargardt¹ (1917).

In Swanzy and Leber's case a large dermoid of the cornea was connected by a short thick pedicle with a further mass which projected outside the lids. At operation the eye was opened, with escape of vitreous. There was no trace of lens or anterior chamber. The abnormal fibrous tissue of the cornea blended with the iris. The presence of the constricted pedicle between the abnormal cornea and the pendulous extra-palpebral mass is explained easily as due to pressure on the base of the tumour by the growing lid folds which nip it. It affords a further piece of evidence of the very early date at which the anomaly arises, *i.e.*, before the lid folds are present (*circa* 18 mm.).

Hanke's case³ is embryologically similar but clinically different as the mass was very small and did not project beyond the lids. The child was 14 years old. The mass of fibro-fatty tissue extended backwards in its upper part. The lens was absent. There were traces of iris especially below, where there was a small space possibly representing the anterior chamber.

Stargardt describes an extremely interesting case, clinically resembling Swanzy and Leber's² in that the tumour was in two parts united by a constricted pedicle. In this case two operations were performed, the tumour only being removed at the first, the rest of the eye subsequently. The iris, lens and anterior chamber were absent. There was no fat in the inside of the eye but the extra-palpebral portion of the tumour was a fibro-lipoma. One extremely interesting feature of the case was the fact that there was a persistent hyaloid running from the optic disc into the posterior surface of the corneal mass. A strand of vascular tissue containing nerve fibrils ran forwards from the anterior end of the hyaloid through both tumours and the intervening pedicle.

This connection of a persistent hyaloid with a fibro-lipoma of the cornea is also noted by Wagenmann⁴ who described in 1889 a case

of fibro-lipoma of the cornea of the size of a hen's egg. At first sight this appears similar to the present case, but it shows the important morphological difference that rudiments of the lens were present, thus retarding the date of origin until after the 10mm. stage at least. Stargardt places this case in the first group but this is debatable in view of the presence of the lens.

The present case resembles that of Stargardt most closely though it is not possible on the material at hand to prove the persistence of the hyaloid artery. The tumour was probably not as large as Stargardt's and only shows a slight constriction round its base, which was embraced by the lids. The condition appears, however, to be of sufficient rarity to warrant its publication.

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THE SURGERY OF THE VITREOUS:

A preliminary paper dealing with an experimental investigation into the Surgery of the Vitreous Body with special reference to its application in cases of Detached Retina, Primary Vitreous Disease, Haemorrhage or Traumatic loss of Vitreous.

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History

The vitreous body, the very origin of which is yet open to speculation^{1,2}, may in common with all living matter undergo changes both physiological and pathological that profoundly influence the vitality and function of its neighbouring structures. Between those delicate alterations of substance that produce the subjective symptoms associated with "muscae volitantes" on the one hand, to the sweeping havoc of a panophthalmitis on the other, lie conditions the ultimate termination of which are largely controlled by the state of the vitreous. A diseased vitreous body is a source of potential danger to sight both directly and indirectly, for in the first instance visual acuity must weaken when a hazy