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APLASIA OF THE PAPILLA AND RETINAL VESSELS WITH A PECULIAR ANOMALY AT THE MACULA IN EYES OTHERWISE NORMAL.

By

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(*With two text-figures.*)

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THE following case seems worthy of record:

Fred R., aged two and one-half years, was brought to the Cornell Dispensary, December 5, 1902. At age of three months it was noticed that child paid no attention to objects waved before the eyes, and since that time has never seemed to see anything. Other senses good. Hearing in particular is keen, and child discriminates tunes and is fond of music. Physical development in general good, although child can neither stand nor walk. Mental development said to be equal to the ordinary. Temper and disposition good.

No illnesses of any kind. No convulsions except one attack during teething at age of one.

Patient when first examined restless, and made continual purposeful movements of head, arms, hands, and legs. When seen at other times was quiet.

Both eyes make searching to-and-fro movements, and besides show a constant, very slight, but rapid nystagmus, mainly vertical in direction.

Apparently no perception of light. Winks at times when light is thrown into eyes, but at other times does not, and seems not to follow the light.

Eyes normal in size. Sclera, cornea, iris, and lens normal. Pupils moderately wide and equal. No light-reaction.

Large floating opacities in vitreous of left eye; none in right.

Discs in both eyes irregularly oval; are of a uniform dirty white, devoid of markings, and look flat and inconspicuous. The papillary vessels are limited to two very slender twigs, arising in each eye from the centre of the disc and running respectively up and down to the border of the disc. Those of the left eye (Fig. 2) cannot be traced beyond the disc; those of the right eye (Fig. 1), which can be followed a little way into the retina, all appear to bend away to the nasal side.

Except for these twigs, *no vessels, either retinal or choroidal, are to be seen in the entire fundus.*

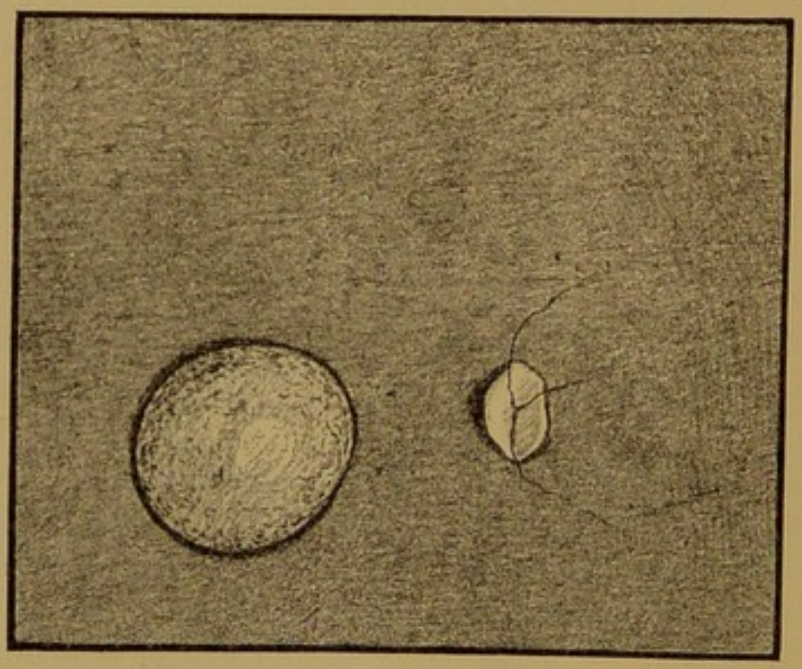

FIG 1.—RIGHT EYE (ERECT IMAGE).

About one papilla-breadth from the disc, to its temporal side, there is in each eye a nearly *circular area, several disc-diameters wide.* The area is bounded by a sharp-cut dark border, so as greatly to resemble in appearance a bubble as seen under the microscope. Within this area the choroidal red is still present, but rather paler than in the surrounding fundus, and there is also a little scattered brownish pigment. There does not seem to be any special difference in level between the floor of the area and the surrounding fundus, although precise estimates cannot be made on account of the constant nystagmus and also because of the absence of any distinctive markings upon which the ophthalmoscope could be accurately focused.

The surrounding fundus, apart from the total absence of vessels, appears perfectly normal.

The refraction, estimated by skiascopy under atropine, was about 0.5 D of myopia.

Here there was evidently *aplasia of the retinal vessels and probably also of the papilla*, which besides being bloodless was misshapen, pale, inconspicuous, and destitute of the usual markings. The appearances of the disc, indeed, were much like those obtaining in a marked post-neuritic atrophy. There were, however, absolutely no other evidences of in-

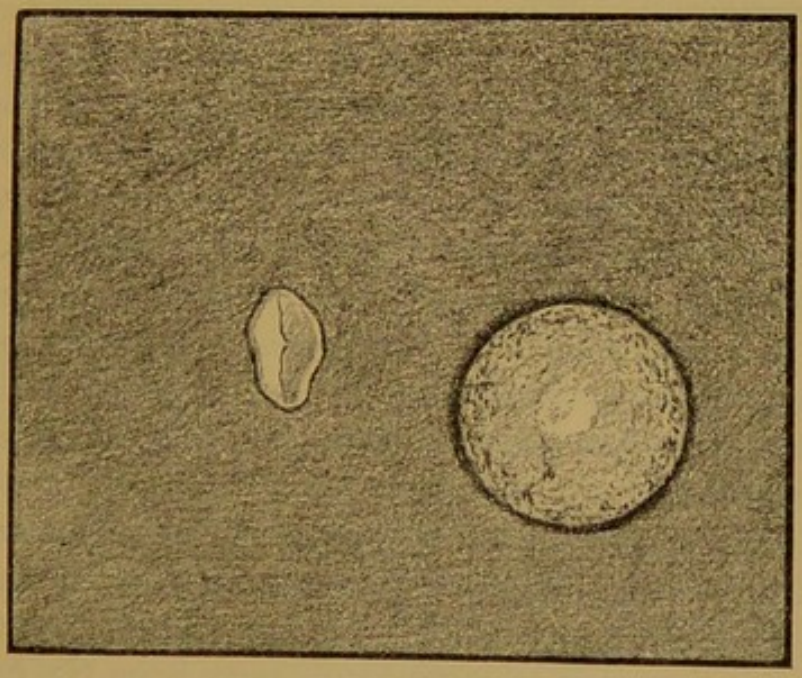

FIG 2.—LEFT EYE (ERECT IMAGE).

flammation. Moreover, according to the authors to be cited later, these congenital anomalies of the retina and nerve are rarely, if ever, due to inflammation, even when the gross appearances are those of an inflammatory process. The changes in the disc in the present case, therefore, seem more likely to be attributable to arrest of development rather than to an antecedent neuritis.

Furthermore, there was at the macula in each eye a large circular area, which gave the appearance of there being *either a defect in the retina* or else an abrupt, *cyst-like, perfectly transparent protrusion of the retina* at this spot. Which was the condition actually present could not be

stated with certainty, owing to the entire absence of retinal vessels or other markings that would have shown the structure and contour of the membranes. There was certainly no protrusion of the choroid, and it is not easy to understand how, without a simultaneous protrusion of the choroid, a cystic protrusion of the retina could have existed for two or three years and maintained its transparency. Yet the sharp black edge of the area, resembling greatly the dark rim of a bubble seen under a microscope, or of the edge of a dislocated lens when seen by transmitted light, strongly suggested the presence of a smooth, transparent, bladder-like prominence.

The case seems to be almost unique in that both the peculiarities described were present in eyes otherwise well formed and in a child of fairly normal development.

Aplasia of the optic nerve is not infrequent in *anencephalia*. In this condition various investigators[1] have found the ganglion-cells wanting in the retina and the nerve-fibres absent in both retina and optic nerve, the latter containing only glia or connective tissue.

Rosenbaum[2] has found a similar condition in *congenital hydrocephalus*.

Van Duyse[3] in a *cyclopean eye* found the optic nerve and papilla replaced by a mass of connective tissue, enclosing numerous vessels and ciliary nerves. There were also, at spots corresponding to what would have been the maculæ in the two component eyes, areas in which the choroid and pigment epithelium were absent, and the retina reduced to the supporting structures only ("coloboma of the macula"). There was furthermore a median inferior coloboma of the choroid; also two lenses; and finally two pupils, each in connection with a coloboma of the iris and ciliary body.

There may be more or less aplasia of the nerve and of the central vessels in *microphthalmus*. Dötsch[4] cites a remark-

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[1] Wahl, Manz, Pétren, and others cited by Rosenbaum (*Ztschr. f. Augenh.*, vii., 2, 1902), who records two additional cases.

[2] *L. c.*

[3] *Arch. d'ophtalmol.*, 1899, pp. 25 etc.

[4] *Arch. f. Ophth.*, 1899 (1), p. 59.

able case, observed in a two-days-old child. The right eye measured 7.5 x 8.0*mm*. In this the nerve was replaced by a thin connective-tissue strand, which contained no nerve-fibres. The retina also was devoid of nerve-fibres, but did contain ganglion-cells, although these were fewer than normal. There were no retinal vessels and no trace of a papilla. The retina showed in places various abnormities of structure; the choroid, ciliary body, and iris were undeveloped; there was a coloboma of the iris and ciliary body; and the cornea was opaque. In the left eye, which measured 8 x 10*mm*, the cornea was opaque and there were other changes, but the papilla, nerve, and retinal vessels were present, and the retina was fairly normal.

The total absence of retinal vessels in the right eye in the case last cited is noteworthy. For, as Dötsch points out, while the central artery is frequently absent in microphthalmus,[1] there is generally a substitute for it, formed by one or more vessels which perforate the sclera or the edge of the lamina cribrosa.

It is to be noted that in none of these cases of congenital anomaly were the changes such as could be attributed to inflammation. Hess[2] in discussing microphthalmus shows that what with the ophthalmoscope, and even at first glance with the microscope, may look like inflammatory products (*e. g.*, pigment deposits, etc.) are not really inflammatory in nature.

In the cases just cited there was in all some obvious and considerable vice of development (anencephalus, cyclocephalus, microphthalmus, congenital hydrocephalus), combined generally with marked abnormities in other parts of the eye beside the nerve and retina. Apart from such abnormities, aplasia of the retinal vessels and of the nerve seems very rare indeed. Interesting anomalies in the formation and appearance of the nerve and papilla, occurring in eyes of otherwise normal aspect, have been described by Purtscher,[3]

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[1] Becker, *Arch. f. Ophth.*, xxxiv., 3; Hess, *ib.*, xxxiv., 3; Bach, *ib.*, xlv., 1.

[2] *L. c.*

[3] *Arch. f. Augenheilk.*, xii. (1883), p. 421.

Eversbusch,[1] Magnus,[2] Derjavine,[3] and others, but these cases have not even a remote resemblance to the one under consideration. I have not yet been able to find a record of an ophthalmoscopic examination of an eye, exteriorly normal, in which the retinal vessels were, as in my case, almost totally absent.

The *second condition* present in this case also seems very rare. It is obviously quite different from the so-called coloboma of the macula, in which the pigment epithelium and choroid are absent, so that the sclera is exposed. The only case that I can find which at all resembles it is one described by Birnbacher.[4]

A man, twenty-one years old, had V = $\frac{6}{36}$ in each eye with − 7.00D. Field normal; no central scotoma. Light-sense normal. Papilla and retinal vessels normal.

In each eye the macula was occupied by a circular structure 1.5 or more papilla-diameters wide. A deep black pigment ring surrounded the figure and sent jagged processes into it. Choroidal vessels crossed it; retinal vessels ran close to it, but did not cross it. The structure, although disc-like in appearance, was evidently conoidal, and, in fact, many *mm* high, for, while its apex (including the choroidal vessels which crossed over it) showed by the ophthalmoscope a refraction of − 8D in one eye and − 7D in the other, the papilla and remaining fundus—even right up to the edges of the prominence—had a refraction of − 25D.

Here there was evidently a finger-like protrusion consisting of both choroid and retina, and possibly also of the sclera as well, although Birnbacher thinks this unlikely on account of the very sudden change in elevation at the margin of the prominence. In any event, the case differed from mine, since in the latter there was no elevation of the choroid and possibly none of the retina, though the appearances strongly suggested a protrusion of the latter.

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[1] *Klin. Monatsbl. f. Augenheilk.*, 1885, p. 1.
[2] *Ibidem*, 1885, p. 43.
[3] *Wiestnik Ophtalmolog.*, Jan.–Feb., 1896, cited in *Arch. d'ophtalmol.*, 1896.
[4] *Arch. f. Augenheilk.*, xv. (1885), p. 159.