PROPTOSIS CAUSED BY LIPOIDOSIS*

BY

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AMONG cases of proptosis, I have met with three in which the causative lesion proved to be a form of lipoid granulomatosis, also known as lipoidosis and xanthomatosis. Two of these cases will be presented in this paper; in the third (Handousa, 1949) the lesion was mainly manifested in the temporal bones simulating double mastoiditis.

The term lipoidosis covers several clinically distinguishable entities of disease resulting from disturbed fat metabolism. Three main varieties are usually described: Gaucher's disease, Niemann-Pick's disease, and Hand-Christian-Schüller disease. They have the common feature of storage of lipoid material in large cells derived from the reticulo-endothelial system. These cells are histologically characteristic and sufficiently diagnostic of lipoidosis. The type of lipoid stored differs in each entity. Kerasin is the main lipoid in Gaucher's disease, phosphatide in Niemann-Pick's disease, and cholesterol and its esters in Hand-Christian-Schüller disease.

Proptosis caused by lipoidosis is very rare and is usually one of the manifestations which may occur early or late in Hand-Christian-Schüller disease. Granulomatous masses forming at the base of the skull in this disease commonly spread and invade the orbit, pressing on the pituitary body and neighbouring structures, thus giving rise to exophthalmos, diabetes insipidus, etc. (MacCormac, 1938). In the case previously published (Handousa, 1949) the lipoid infiltration was a generalized process in which the orbital involvement was late.

The possibility of a localized process of lipoidosis in the soft tissues of the orbit without any skeletal, visceral, or skin manifestations is demonstrated by the two cases here presented.

Case Reports

Case 1. S. S., male, aged 40 (Fig. 1), complained of right proptosis of about 2 years' duration, gradually and slowly increasing without any pain, headache, or change in vision. No history of trauma or syphilis. No diplopia.

Examination.—The right globe was bulging forwards and slightly downwards with limitation of upward movements. Conjunctiva clear; vision in the right eye 6/6.

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Palpation revealed a cystic swelling lying in relation to the roof of the orbit and extending above the eyeball from the medial to the lateral side, with the tendon of the superior oblique muscle stretched anterior to it. At the outer limit of the swelling, the orbital edge was irregularly thickened.

The left eyeball also appeared slightly proptosed, but there was no palpable swelling in the orbit and no limitation of ocular movement. Vision in the left eye 6/6. Fields of vision normal. Nose and sinuses showed no related abnormality.

Laboratory Findings.—Wassermann reaction negative; basal metabolic rate -17 per cent. Urine and stools normal. General clinical examination revealed no abnormality.

Blood:

Total red-cell count 5,100,000, haemoglobin 92 per cent.

Total leucocyte count 4,200 (polymorphs 46 per cent., lymphocytes 42 per cent., monocytes 10 per cent., eosinophils 2 per cent.).

Radiological Examination.—This revealed a large defect in the orbital roof (Figs 2 and 3). No other bony lesion was discovered in the skull or other bones.

FIG. 1.—Case 1, right proptosis caused by a localized xanthomatous mass.

FIG. 2.—Case 1, radiograph of right orbit, showing ? defect in bony roof. Exploration revealed the apparent defect on the right side to be merely an upward bulge in the orbital roof forming the bed of the mass.

FIG. 3.—Case 1, radiograph of left orbit (cf. Fig. 2).

Exploration.—A fairly large but localized soft mass was found mainly in relation to the outer part of the orbital roof. It was adherent to the mass of the muscle cone with the tendon of the superior oblique muscle spread anterior to it. It could be dissected out completely. No trace of the lacrimal gland was found. The part of the orbital roof forming the bed of the mass was pushed up intra-cranially, but was still intact, and the bone surrounding this bed was irregularly thickened. Figs 4 and 5 give a diagrammatic representation of the position of the mass in the orbit.

FIGS 4 and 5.—Case 1, diagrammatic representations of xanthomatous masses *in situ*.

On section, the cut surface of the mass was dark brown in colour and the fluid which oozed out was full of minute shining particles which proved to be cholesterol crystals.

Histological Examination.—The mass proved to be xanthomatous in nature (Fig. 6).

FIG. 6.—Case 1, photomicrograph showing (1) needle-shaped spaces of dissolved cholesterol crystals, and (2) groups of lipoid foam cells.

Follow-up.—The patient had an uninterrupted recovery, and the proptosis has disappeared. He has been seen regularly since the operation (for 7 years) and is in perfect health.

Case 2. H. M. T., boy, aged 4 (Fig. 7), complained of progressive exophthalmos of the left eye of one year's duration, associated during the last month with ptosis of the left upper eyelid.

The condition developed insidiously and painlessly without any fever or constitutional disturbances; the parents state that when one year old the child had ophthalmia which left him with a nebula on the left cornea.

There was no history of trauma or syphilis, and the child had two brothers in good health and free from similar complaints.

Examination.—The left eyeball was proptosed forwards, downwards, and medially, with limited movements in an upward and downward direction, and ptosis of the upper eyelid. The conjunctiva was congested and showed distinct ecchymosis in its lateral part. There was a nebula on the lower part of the left cornea. On palpation, a soft mass could be felt in the upper and outer quadrant of the orbit in the region of the lacrimal fossa. The mass was not tender, and was not adherent to the skin above it; but its extent in depth could not be discovered. The orbital margin was intact all round. The anterior chamber was free and of normal depth; fundus normal.

No abnormality was detected in the right eye, and the neck and pre-auricular regions were free of palpable glands. Except for the presence of adenoids, the nose, nasopharynx, throat, and ears were normal. The general condition was quite good, pulse and temperature were normal. No clinical or radiological abnormality was discovered in the lungs, heart, or abdomen. The size of the liver was within normal limits and the spleen was not palpable. The skin was free of xanthomatous masses.

Laboratory Findings

Urine:

Acid, Sp. gr. 1016.
Albumen-traces.
Pus cells + Red blood corpuscles +.
Bence-Jones reaction—negative.

Blood:

Red-cell count 5,000,000, haemoglobin 90 per cent.
Leucocyte count 12,700 (polymorphs 48 per cent., lymphocytes 43 per cent., monocytes 6 per cent., eosinophils 3 per cent.).
Blood cholesterol 150 mg. Wassermann reaction negative.

Sternal puncture.—Total count 120,000.

(Basophils 10 per cent., staff nucleated 17 per cent., eosinophils 11 per cent., segmented 30 per cent., juveniles 0 per cent., promyelocytes 8 per cent., myeloblasts 8 per cent., lymphocytes 12 per cent., monocytes 4 per cent.).

Radiological Examination.—The left orbit compared with the right was dilated, but its margin was free of erosion. The orbital mass threw a shadow on the orbital cavity covering its upper and outer margins (Fig. 8, overleaf). No radiological lesion was discovered in the flat or long bones.

Exploration.—A large mass, brown in colour, was seen lying between the muscle cone and the orbital wall, extending back almost to the optic foramen and infiltrating all the neighbouring structures. The lacrimal gland could not be made out, but the bony orbital wall looked and felt uneroded. In view of this infiltration I removed only a piece of the mass for biopsy.

Histological Examination.—A mass of lymphoid tissue presenting marked hyperplasia of its reticulo-endothelial cells with numerous groups of large, pale, vacuolated, lipid-laden cells, which gave a mild positive reaction for fat in frozen sections stained by the Sudan III method; the appearances were compatible with lipid histiocytosis, most probably of the Hand-Christian-Schüller type (Fig. 9, overleaf).

Treatment.—The patient is receiving deep x-ray therapy.

FIG. 7.—Case 2, left proptosis and ptosis, caused by xanthomatous mass in orbital space.

DISCUSSION

In these two cases, proptosis was the only complaint and was proved to be caused by a xanthomatous intra-orbital mass.

FIG. 8.—Case 2, radiograph showing primary dilatation of left orbit and shadow of lipoid mass superimposed upon it.

FIG. 9.—Case 2, photomicrograph of section of lipoid mass showing (1) lipoid-laden foam cells, and (2) eosinophil cells.

The mass was large and infiltrative in Case 2, but localized in Case 1. In both cases the mass was found mainly in the upper and outer quadrant of the orbit and the evidence was in favour of origin in the intra-orbital soft tissue. Radiological study, exploration, and histological examination gave

no indication that the mass originated from bone. The lacrimal gland was absent on exploration, having most probably disappeared as a result of pressure atrophy. The serial sections examined histologically did not reveal any evidence of lacrimal gland tissue in the mass. There was no evidence of any similar process of disease in other parts of the body.

Proptosis is one of the manifestations of Hand-Christian-Schüller disease and commonly results from spread to the orbit of the lipoid granulomata forming at the base of the skull. *These two cases demonstrate that proptosis can also be caused by a similar process originating in and limited to the soft tissues of the orbit.*

SUMMARY

Two cases of proptosis caused by a xanthomatous intra-orbital mass are described, and the aetiology is discussed in detail.

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