CASE NOTES

NEONATAL NEUROFIBROMATOSIS*

BY

J. GIBSON MOORE

Edgware Hospital, Middlesex

VON RECKLINGHAUSEN'S neurofibromatosis is a common disease with varying manifestations but its primary appearance in the eye or lids is uncommon.

Worster-Drought, Dickson, and McMenemey (1937) recommended that the term "von Recklinghausen's disease" should be confined to cases exhibiting only cutaneous lesions and suggested that the term neurofibromatosis should be used when there was, in addition, evidence of central lesions. There is still, however, some confusion in classification, as some authors state that neurofibromatous lesions are produced by diffuse overgrowth of the nerve sheaths and their supporting connective tissue, but make a distinction between these tumours and those arising from the neuro-ectodermal sheath of Schwann (schwannoma or neurilemmoma).

It is probable that there is a distinct nerve-sheath tumour, usually solitary, which can be classified as a schwannoma (Skeoch, 1956). This may be solitary and benign, but solitary tumours have also been described showing the changes of neurofibromatosis and devoid of neurilemma structure.

On the other hand generalized cases of neurofibromatosis may develop characteristics of neurolemmomas, and it seems likely the nerve-sheath element and the cells of the sheath of Schwann are jointly involved in such tumours, the pathological appearance and classification of the tumour depending upon which structure predominates.

Case Report

A **child** was born with an enormous lump in the region of the left orbit (Figs 1 and 2, opposite). The maternal history was negative and the birth normal.

Examination.—The lump was red in appearance and solid to palpation, and transillumination was negative. There was no apparent covering of skin although there were small veins coursing over its surface. A crescentic area distal to the tumour gives a suggestion of a lower lid, although this was by no means obvious at the time (Fig. 2).

There was no sign of the eye itself and the possibility of its destruction by a meningioma was considered.

X-ray examination showed a normal orbit with no sign of bony erosion or abnormality of the optic foramina. A blood count showed a hypochromic anaemia but no abnormality of the cells.

* Received for publication November 27, 1961.

FIG. 1.—Appearance at birth, side view.

FIG. 2.—Appearance at birth, front view.

There were considerable domestic difficulties to contend with. The mother on seeing the child became upset almost to the point of dementia and refused to have anything to do with it. She announced her intention of abandoning the baby and leaving it in the care of the hospital. It was therefore decided to operate even though the child was only 14 days old.

Operation.—The lump was removed, and the tumour was solid and “gritty” when sectioned. There was very free bleeding at the time but a reasonably satisfactory cosmetic removal was obtained. It became clear that the margin of the upper lid was actually present and that an apparently intact eye was present under the lump.

Pathological Report.—“The structure and staining reactions of the tumour suggest that it is a simple fibroma (Fig. 3), although the possibility of neurofibroma cannot be excluded and I suppose that in the latter case recurrence is more likely if the removal is incomplete. Another possibility to be excluded is a meningioma eroding through the orbital bone.”

Progress.—Recovery was uneventful and a year later the position was as in Fig. 4. There is considerable improvement although the cosmetic position is by no means perfect.

FIG. 4.—Appearance at 12 months.

FIG. 3.—Histological section of orbital tumour.

The eye itself is intact with normal-sized cornea and a healthy iris. The pupil is active and the ocular movements full although a convergent squint is present. The retina is normal except for medullated nerve fibres above the disc. It is interesting that eye-lashes have now grown on both lids.

A second small lump found on the child's back was also removed, and the pathological report was as follows:

"Section shows a small mass consisting of collagen fibres and spindle cells with a somewhat whorled appearance. The appearances are compatible with neurofibroma" (Fig. 5).

FIG. 5.—Histological section of tumour excised from the back.

The mother is very happy at the present time and is well reconciled to the position. She understands that further surgery can be attempted to improve the child's appearance.

Summary

A case of neurofibromatosis presented at birth with a large lump apparently occupying the orbit. The pathology of this condition is briefly discussed. Although the case produced no new facts, it is presented as an unusual clinical phenomenon.

I wish to acknowledge the help and pathological reports of Dr. J. L. Hamilton-Paterson, Senior Pathologist at Edgware Hospital.

REFERENCES

- SKEOCH, H. (1956). *Brit. J. Ophthalm.*, **40**, 562.
 WORSTER-DROUGHT, C., DICKSON, W. E. CARNEGIE, and McMENEMEY, W. H. (1937). *Brain*, **60**, 85.