HEMICRANIAL APLASIA WITH
PULSATING EXOPHTHALMOS
AN UNUSUAL MANIFESTATION OF VON RECKLINGHAUSEN'S DISEASE

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WHEN osseous defects are present in patients suffering from von Recklinghausen's disease, it is usually assumed that these defects are the result of local bone destruction produced by contiguous expanding or invading neoplasms. However, it is apparent, upon review of the literature, that in addition to the cutaneous and nervous-system manifestations of this curious disease, primary disorders of bone are frequently present. After a review of records and roentgenograms of 127 patients with von Recklinghausen's disease, Holt and Wright estimated that 29 per cent of the affected group had some form of skeletal defect. In attempting to classify the types of bone changes seen, it was their conclusion that congenital anomalies and scoliosis were the most common abnormalities. However, bowing and pseudoarthrosis, localized overgrowth or atrophy, and bone destruction caused by expanding contiguous neoplasms were also seen.

In a monograph on this subject, it was the feeling of Crowe et al. that only rarely does a patient with generalized neurofibromatosis fail to show some evidence of skeletal involvement. It thus appears to be increasingly evident that von Recklinghausen's disease is a diffuse, probably genetically established disorder with its effects often manifest in the skin, peripheral and central nervous systems, and in bone.

A characteristic but relatively rare abnormality of the cranial vault in patients with neurofibromatosis has been reported by several authors in the past. In the patients described, there has been a unilateral agenesis of the sphenoid bone and posterior orbital bone often associated with atrophy of the ethmoid sinuses and outward bulging of the temporal bone on the affected side. The skeletal defect in the orbit, which results in these patients, permits a unilateral pulsating exophthalmos to develop as a consequence of direct transmission of intracranial pulsations to the globe of the eye. The fact that Petit-Dutaillis et al. in 1949, after a careful survey of the international literature, were able to find only 37 cases of this syndrome in addition to adding 2 cases of their own would attest to the fact that this syndrome is either extremely rare or is not sufficiently recognized.

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During the past year we have been able to study 4 patients who have demonstrated this abnormality. In 2 of the patients a diagnosis of congenital glaucoma was made. The affected eye was removed and a prosthesis was inserted in each case. As might be expected, the defect in the posterior orbit continued to transmit intracranial pulsations and both patients exhibited pulsations of their prostheses synchronous with cardiac systole.

In addition to the usual radiographs of the skull, tomograms in anteroposterior and lateral projections have been obtained in all 4 cases. Contrast radiographic studies of the brain were carried out in 2 of the 4 patients.

The histories of these 4 patients are presented in an effort to make clinicians and radiologists aware of an unusual but characteristic manifestation of neurofibromatosis.

CASE REPORTS

Case 1. MGH #413361. B.A.G., a 28-year-old white, married female, was first seen by the Neurosurgical Service of the Massachusetts General Hospital in February, 1958. Her presenting symptoms were intermittent attacks of bitemporal headache of about 6 months' duration. She had previously been followed by the Ophthalmological Department of the Massachusetts Eye and Ear Infirmary since 1943.

At the age of 2 months, it had been noted that her left eye protruded. A diagnosis of "congenital glaucoma" was made. The left eye continued to be protuberant and in 1936, when the patient was 6 years old, the left eye was removed at an outside hospital. A prosthesis was inserted following orbital exenteration.

In 1943, when the patient was 13 years old, she presented herself to the Massachusetts Eye and Ear Infirmary with ptosis of her left upper lid. At this time it was noted that her prosthesis did not "fit well" but no mention was made of any pulsations of the prosthesis. Nodular excrescences were felt in the ptotic left lid and these were subsequently removed. The histologic diagnosis of these nodules was neurofibromata.

In 1945 she was again admitted to the Massachusetts Eye and Ear Infirmary because of increasing proptosis of her prosthesis. On this occasion more nodules of neurofibromatous tissue were removed from the left orbit. There was no extension of the neoplasm beyond the orbital confines. Radiograms of the skull on that occasion were reported as showing "marked expansion of the left orbit. The roof and apex of the orbit were thinned out. There was no evidence of bone erosion."

In 1948 she was first seen by the Dermatology Department of the Massachusetts General Hospital because of multiple fibromas of the skin and café-au-lait spots. A diagnosis of generalized von Recklinghausen's disease was made at that time.

In October, 1957 the patient returned to the Eye Department of the Massachusetts Eye and Ear Infirmary because of intermittent attacks of bitemporal headaches. Radiograms in October, 1957 revealed "enlargement of left orbit with evidence of previous exenteration of the left eye. The lesser and greater wings of the sphenoid bone on the left side are gone. There is evidence of destruction of the sella turcica." Because of the bone destruction and headache, it was felt that the patient was harboring an expanding neoplasm—probably a neurofibroma—in the floor of the middle cranial fossa with erosion of the posterior wall of the left orbit. From Nov. 12 to Dec. 5, 1957 she had a total of 3000 r (tumor dose) given in divided doses by the 2 mev. unit to the middle cranial fossa on the left side. Irradiation pro-