Familial Pheochromocytoma with Ependymoma of the Spinal Cord

Case Report and Review of the Literature*

KARL O. VON HAGEN, M.D., AND HOWARD S. BARROWS, M.D.

Department of Neurology, University of Southern California, School of Medicine, and Neuromedicine Service, Los Angeles County General Hospital, Los Angeles, California

Multiple neurofibromatosis occurs in 5–25 per cent of patients with pheochromocytomas.1,3,4 The first recorded case of pheochromocytoma with a meningioma of the foramen magnum without evidence of neurofibromatosis was reported by Greenhouse in 1961.5 He felt that this was more than a fortuitous relationship, since meningiomas are a frequent accompaniment of neurofibromatosis. His review of the literature emphasized the relationship of pheochromocytoma to the other manifestations of multiple neurofibromatosis. He felt that his case might provide further evidence to justify the concept that the neurocutaneous syndromes—multiple neurofibromatosis (von Recklinghausen's disease), tuberous sclerosis (Bourneville's disease), meningofacial angiomatosis (Sturge-Weber's disease) and cerebelloretinal hemangioblastomatosis (von Hippel-Lindau's disease)—are connected with pheochromocytomas. If so, one would expect tumors of other neural elements to occur in combination with pheochromocytomas as they do with the other neurocutaneous syndromes.

Recently, a girl with familial pheochromocytoma and an associated ependymoma of the spinal cord was studied on the neurologic service of the Los Angeles County General Hospital. This patient showed no evidence of multiple neurofibromatosis despite the notable association of ependymomas with multiple neurofibromatosis.6 We are presenting her case and shall discuss its possible significance.

Report of Case

The patient's brother was the first to present himself to the Los Angeles County General Hospital. Prior to his admission he had recurrent episodes of excessive sweating since the age of 15. On occasion these were so severe that he had cramps in his muscles and encrustations of salt in his shirt. In January 1960, at the age of 20, he was admitted in shock with severe dehydration following an attempted 8-day drive through the desert. He responded to appropriate symptomatic therapy and was found to have markedly elevated urinary catecholamines. Surgical explorations revealed a left adrenal pheochromocytoma, a right adrenal pheochromocytoma, a left ectopic pheochromocytoma and a right ectopic pheochromocytoma. These were all removed. Dr. Shannon Brunjes screened 14 members of the boy's family in three generations for urinary catecholamines. Significantly elevated levels were demonstrated in the boy's father and in one sister of a sibling of four. This sister, our patient, is a 13-year-old white female.

She was admitted to the Los Angeles County General Hospital in March 1961 for further studies. Her only complaint at that time was intermittent sweating, flushing and swelling of her hands and to a lesser degree her feet. An elevated blood pressure had never been demonstrated.

Examination and numerous laboratory tests, including intravenous urograms and retroperitoneal pneumography, revealed no abnormalities.

Operation. An exploratory laparotomy in April 1961 revealed a pheochromocytoma on the right adrenal gland which was removed by a partial adrenalectomy. The left adrenal gland was visualized as normal and no ectopic tumors were noted.

Histologic Report (Dr. Weldon Bullock). Sections of this tumor were typical of pheochromocytoma (Fig. 1).

Course. The patient complained of severe pain in the right side of her neck on the 4th postoperative day. This complaint may have been present in a mild form prior to operation. Roentgenograms of the cervical spine, right scapula and shoulder were taken and reported as being within
normal limits. Her postoperative urinary catecholamines were normal.

During visits to the clinic the patient was noted to have flushing and moisture of her hands and complaints of ever worsening pain in the neck. This pain became progressively worse despite temporary relief from physical therapy. She was unable to sleep, except in a near-sitting position, by September 1961. Whenever the pain in the neck was severe, standing and walking would relieve it to some degree. She became aware of weakness in both arms, more marked on the left, during the few months prior to her second admission to the neurologic service in November 1961. Coincident with this was an increased tendency to drop objects. She became so disabled prior to admission that she was unable to successfully button and unbutton her clothes or dress herself.

Examination on 2nd admission revealed an alert, cooperative, thin and anxious young girl. There was obvious atrophy of the muscles in the left upper extremity proximally and in the shoulder. She had moist, mottled, bluish discoloration of both hands with minimal puffiness. Blood pressure was 120/80, and pulse was regular, with a rate of 100. Her face had an erythematous hue over both malar areas. The feet were cold and slightly erythematous.

Neurologic examination revealed a thin girl with markedly reduced masses of muscles generally. She held her neck very rigidly in an extended position. She was unable to tolerate movement of the neck in any direction. There was marked suboccipital tenderness to palpation bilaterally. She evidenced obvious atrophy of the muscles of the shoulder bilaterally, most marked on the left. She was weak in all the muscles tested in both upper extremities. The weakness was most marked in the proximal left arm. She was unable to abduct the arm or flex the elbow against gravity. An early flexor contracture of the middle finger of the left hand was seen. The triceps reflex was absent in the left upper extremity and hyperactive in the right upper extremity. The finger reflexes were not active in the fingers but a marked reflexogenic spread could be seen in the muscles of the shoulder bilaterally. Percussion of the groups of muscles over the shoulders showed increased irritability. The deep tendon reflexes were hyperactive in both lower extremities. Plantar stimulation of the right foot produced a questionable extensor response. Response on the left was flexor. The patient evidenced marked stereanesthesia in the left hand with only slight loss to sensation of a pin in the fingertips of both hands. Sense of position was absent in the left arm, hand and ankle. There was mild cerebellar dysmetria in both hands. The cranial nerves were normal including the fundi. Her radial pulses were readily obliterated with abduction of the shoulders. Roentgenograms of the skull were unremarkable. Roentgenograms of the cervical spine revealed a questionable change when compared to those taken at the onset of her difficulty 7 months previously. This consisted of