STURGE'S DISEASE

BY

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(WITH SPECIAL PLATE)

Rare diseases often acquire a considerable variety of names, and the congenital abnormality about to be described is no exception. It is called "naevoid amentia," Sturge's, Kalischer's, Weber's, Krabbe's, Dimitri's, or Brushfield-Wyatt disease, and not uncommonly it goes under a combination of one or more of the above names.

We agree with Parkes Weber (1936), who, writing under the heading of "Nova et Vetera," modestly disclaims the title to the naming of this condition and suggests that it should be called Sturge's disease. It is true that Schirmer (1860) first noted coexistence of facial naevus with congenital glaucoma, but it was Sturge (1879) who, describing a 6½-year-old girl with an extensive right-sided facial naevus, right-sided buphthalmos, and epileptic fits beginning in the left hand, concluded that the right side of the brain was also involved in the naevoid condition. Sturge maintained his opinion in spite of the adverse criticism of leading contemporary medical men, and the truth of his contention was confirmed by Kalischer (1897), who published the first necropsy findings in a similar case. Subsequently Parkes Weber (1922) showed by means of radiographs that calcification of the brain was present on the same side as the naevus. Full reviews are given by Schiötz (1935), who analysed eighty-nine cases; by Greenwald and Koota (1936), who tabulate eighty-two records; and by Bergstrand, Olivecrona, and Tönnis (1936), who were able to count 108 cases. In many instances confirmation of the brain changes was obtained at necropsy or operation.

Aetiology

Sturge's disease is congenital in origin, and it is permissible to suggest that, by analogy with other comparable disorders, like Recklinghausen's neurofibromatosis, tuberous sclerosis of the brain, or Lindau's disease, it may be hereditary in nature. No definite conclusions can be reached, however, because of lack of relevant records. Only some authors mention that no similar condition was observed in other members of the family; but nowhere is any reference to consanguinity given. There are a few observations nevertheless which bear out the hereditary theory and suggest that a dominant factor may be operative: Oppenheim (1913) found that the mother of a 13-year-old boy with a pial angioma, hemiplegia, and Jacksonian epilepsy herself had a cavernous angioma of the lip; Kufs (1928) gave details of a man of 81 with angiomata of face and brain, and inferred from the previous history of one of the daughters that she had an angioma of the pons; and Geyelin and Penfield (1929) described calcification of brain and epilepsy in a man and his four children.

Sturge's disease has been reported in many parts of the world, but mostly in Europe. The total number of cases we were able to count, including our own case, is 145. With regards to sex there is a slight preponderance of males.

Pathology

The principal features are: (1) A cavernous naevus, the colour of a port-wine stain, in the distribution of one or more branches of the fifth cranial nerve, extending occasionally to areas of skin in other parts of the body and rarely involving the mucous membranes. (2) Vascular changes in the eye on the side of the naevus. (3) Angiomatous changes in vessels of the pia. (4) More or less marked changes in the brain, such as hypoplasia, atrophy, and calcification confined to one side of the cerebrum—the same side as the facial naevus, in cases in which this too is present.

The changes in the pial vessels may vary from a slight telangiectasia to a racemose venous angioma. The calcification, which is actually in the cortex and the subcortical layers, is partly perivascular and partly a deposition of free calcium granules in the brain substance. In the calcified areas the number of nerve cells is greatly reduced and glial tissue relatively increased. These findings suggest that the brain changes are more likely to be part of a local agenesis than a consequence of the superficial naevoid condition.

Clinical Manifestations

The facial and other cutaneous naevi are a prominent sign and are present at birth, but occasionally the naevus may be so inconspicuous as to escape notice. The naevus is usually cavernous and flat, but frequently the skin over it is hypertrophied and raised, or even rough and nodular, partaking in an extreme instance (Greig, 1922) of the character of a sebaceous adenoma. The cerebral symptoms come on usually early in life, often before the end of the first decade. They correspond to the extent and position of the brain changes, and usually consist in either Jacksonian or generalized epileptic seizures, hemiplegia, hemiatrophy, and in a lesser or greater degree of amentia. The Jacksonian fits, hemiplegia, and hemiatrophy are, of course, on the side opposite to that of the facial naevus and cerebral calcification, and the same applies to the hemianaesthesia and homonymous hemianopia which are often observed. Some instances of asymmetry of the skull, tendency to adiposity, and sexual hypoplasia are noted. Glaucoma and exophthalmos on the same side as the facial naevus are quoted in about a third of all records. The glaucoma is thought to be due to venous stasis, and when it is present the fundal veins may show naevoid changes. Dilatation of scleral and conjunctival vessels and heterochromia iridis may also be observed. Even in the absence of glaucoma deep cupping of the optic disk may be seen. Papilloedema practically never occurs in Sturge's disease.

The radiological appearances of the skull are very striking, especially in cases where extensive calcification of the brain has taken place. The calcified areas are confined to the affected half of the cerebrum, which is usually smaller than the sound companion. This may be inferred from a plain radiograph in which the outlines of the diseased half of the cerebrum can often be clearly seen, or confirmed by means of a ventriculogram. The cerebellum is not involved. The shadows thrown by the calcium in the cortex are sinuous, and where the rays strike a sulcus at about a right angle two lines running 1 to 2 mm. apart can be seen. The commonest site of calcification is the occipital cortex. Occasionally, instead of or in addition to the usual surface wavy lines, single or multiple strictly demarcated subcortical shadows may be demonstrated.

Diagnosis and Prognosis

Where we have all or most of the components of the syndrome the diagnosis is easy. Occasionally, however, deviations occur from the classical picture, and an important sign—for example, buphthalmos or even naevus—may be missing altogether, or else the cerebral signs may occur on the same side as the naevus. Other minor variations, such as bilateral involvement of the eyes, are also described. It is important to bear the condition in mind in all cases of idiopathic epilepsy, especially if Jacksonian in type, and even in the absence of facial naevus to take a routine radiograph of the skull. Distinction from the arteriovenous type of naevus is drawn by the absence of vascular bruit.

The immediate outlook for life is good. In milder cases a quite useful existence may be expected, but in the more complete and severe forms mental changes are inevitable and sooner or later lead the affected individual to a mental institution. Survival past early middle age is relatively uncommon.

Treatment

The nature of the condition precludes as a rule anything but palliative measures, and this explains why comparatively few patients had any active treatment. The latter includes attempts at removal of the diseased areas of brain, ligation of the common carotid artery, tying or cauterizing the vessels entering into the formation of the pial naevus, freeing of meningeal adhesions, or simple decompression. Good results have been claimed in a few instances for x-ray or radium therapy. Occasionally two or more of the above measures were tried. Failures, including a high mortality rate in the operated cases, appear to be as frequent as successes, and even where amelioration is claimed it may be only temporary. In assessing the value of any treatment it is also important to remember that spontaneous remissions of symptoms are fairly frequent.

Case Record

A man aged 42 was admitted to Erdington House on November 16, 1937, because his brother could no longer afford to look after him. His parents were not related, had both been healthy, and lived till over 80. The patient is fifth in a family of six. One sister died at the age of 30 in a confinement. The rest are healthy. There is no history of naevi or epilepsy in the family.

The naevus on his face was noted at birth, which was normal and at full time. Soon afterwards weakness of the right arm and leg was discovered. He was backward in walking, talking, etc., and was never at school. Since infancy the patient had slight attacks of loss of consciousness. He would go into what appeared like a faint, perhaps slide off his chair, lie without twitching or tremor for a few seconds, and then come round as quickly. Before and during these seizures the facial naevus would become darker. Thundery weather would often be a precipitating factor, and before the "turns" he would usually suffer from left-sided headaches. At first the attacks would occur about once in two to three months; later they became fewer and less pronounced, and now he has been free from them for the last four years. His general health has always been good.

The present mental state of the patient is that of a feeble-minded individual of medium grade. He has full control over his sphincters, is well behaved and co-operative, but incapable of doing any useful work. Physically he is underdeveloped—only 4 ft. 10½ inches tall and weighing 5 st. 10½ lb. The external genitalia appear normal. A bluish-red naevus occupies most of the first and second divisions of the left fifth cranial nerve and extends posteriorly as far as the lambda

(Plate, Fig. 4). The skin over the naevus is hypertrophied and roughened and a few sebaceous glands are very prominent. The circumference of the skull is 20 inches, and from ear to ear over the vertex measures 10½ inches. The left side of the skull is smaller than the right. The forehead is shallow and sloping and the teeth prominent. There is a slight degree of scoliosis in the thoracic region, with the convexity to the left. The eyes are not prominent and show no heterochromia or increase in tension. The pupils react well to light and accommodation, the left being slightly oval. The right fundus appears normal, whereas the left optic disk shows an extreme degree of cupping. Neither eye reveals naevoid changes. Right homonymous hemianopia can be demonstrated. The right arm and leg show some atrophy as compared with the left. The posture, diminution of power, increase in tone, and gait are typical of a moderate right-sided hemiplegia. Weakness of the right lower face is only slight and speech is not affected. All deep reflexes on the right side are markedly exaggerated, and patellar and ankle clonus can be elicited on that side. All abdominal reflexes are present equally. The right plantar is extensor, the left flexor. Sensation is difficult to assess accurately on account of dull mentality, but there appears to be some blunting in superficial sensibility in the right side of the body. Deep sensation is not affected. There is no evidence of cerebellar involvement. Examination of the cerebrospinal fluid showed: pressure normal, cells less than 1 lymphocyte per c.mm., Lange 0001121000, protein 30 mg. per 100 c.cm. The Wassermann reaction was negative, and the heart and lungs were normal. The blood pressure was 110/80. Radiographs (Figs. 1, 2, and 3) show calcification of the left cerebral cortex, most marked in the occipital region. It can also be seen that the left cerebral hemisphere is much smaller than the right.

Summary

A description is given of the main features of Sturge's disease, which are: facial and pial naevi, calcification of brain substance, epilepsy, mental retardation, and glaucoma.

A personal case, showing nearly all the classical manifestations of the syndrome, is recorded.

REFERENCES

- Bergstrand, H., Olivecrona, H., and Tönnis, W. (1936). *Gefäßmissbildungen und Gefäßgeschwülste des Gehirns*, Georg Thieme, Leipzig.
- Geyelin, H. R., and Penfield, W. (1929). *Arch. Neurol.*, **21**, 1020.
- Greenwald, H. M., and Koota, J. (1936). *Amer. J. Dis. Child.*, **51**, 868.
- Greig, D. M. (1922). *Edinb. med. J.*, **28**, 105.
- Kalischer, S. (1897). *Berl. klin. Wschr.*, **34**, 1059.
- Kufs, H. (1928). *Z. ges. Neurol. Psychiat.*, **113**, 651.
- Oppenheim, H. (1913). *Neurol. Zbl.*, **32**, 3.
- Schiötz, E. H. (1935). *Acta Psychiat.*, **10**, 683.
- Schirmer, R. (1860). *Arch. f. Ophthal.*, **7**, 119.
- Sturge, W. A. (1879). *Clin. Soc. Trans.*, **12**, 162.
- Weber, F. P. (1922). *J. Neurol. Psychiat.*, **3**, 134.
- (1936). *British Medical Journal*, **1**, 708.

J. H. Lichtenbelt (*Nederl. Tijdschr. Geneesk.*, 1939, **83**, 1340), who records an illustrative case, states that spontaneous rupture of the stomach is very rare, especially in children, and the prognosis is bad. In only a few cases has recovery taken place after operation. In traumatic rupture the prognosis is better, and recovery has even taken place spontaneously due to the formation of adhesions. The present case was that of a boy, aged 14, who developed rupture of the stomach as the result of fermentation and accumulation of gas in the organ. The abdomen was very much distended, and the blood vessels were compressed, so that there was no circulation in the legs and the boy suffered severe pain. Death took place shortly after admission to hospital, so that no operation could be performed. An hour before death extensive subcutaneous emphysema developed. The condition showed some resemblance to the meteorism which occurs in cattle.

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FIG. 1.—Right lateral view of skull showing calcification of cerebral cortex. Note that calcification is best marked in the occipital region, and double contours represent some of the sulci.

FIG. 2.—Postero-anterior view of skull demonstrating the marked hemiatrophy of the brain and some atrophy of the skull on the same side.

FIG. 3.—Antero-posterior view of skull demonstrating the marked hemiatrophy of the brain and some atrophy of the skull on the same side.

FIG. 4.—Extent of naevus is only partly shown; the rest is covered by hair. Note posture and wasting of right upper limb.