STURGE - WEBER SYNDROME

THESIS

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By

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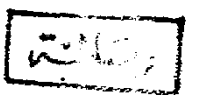

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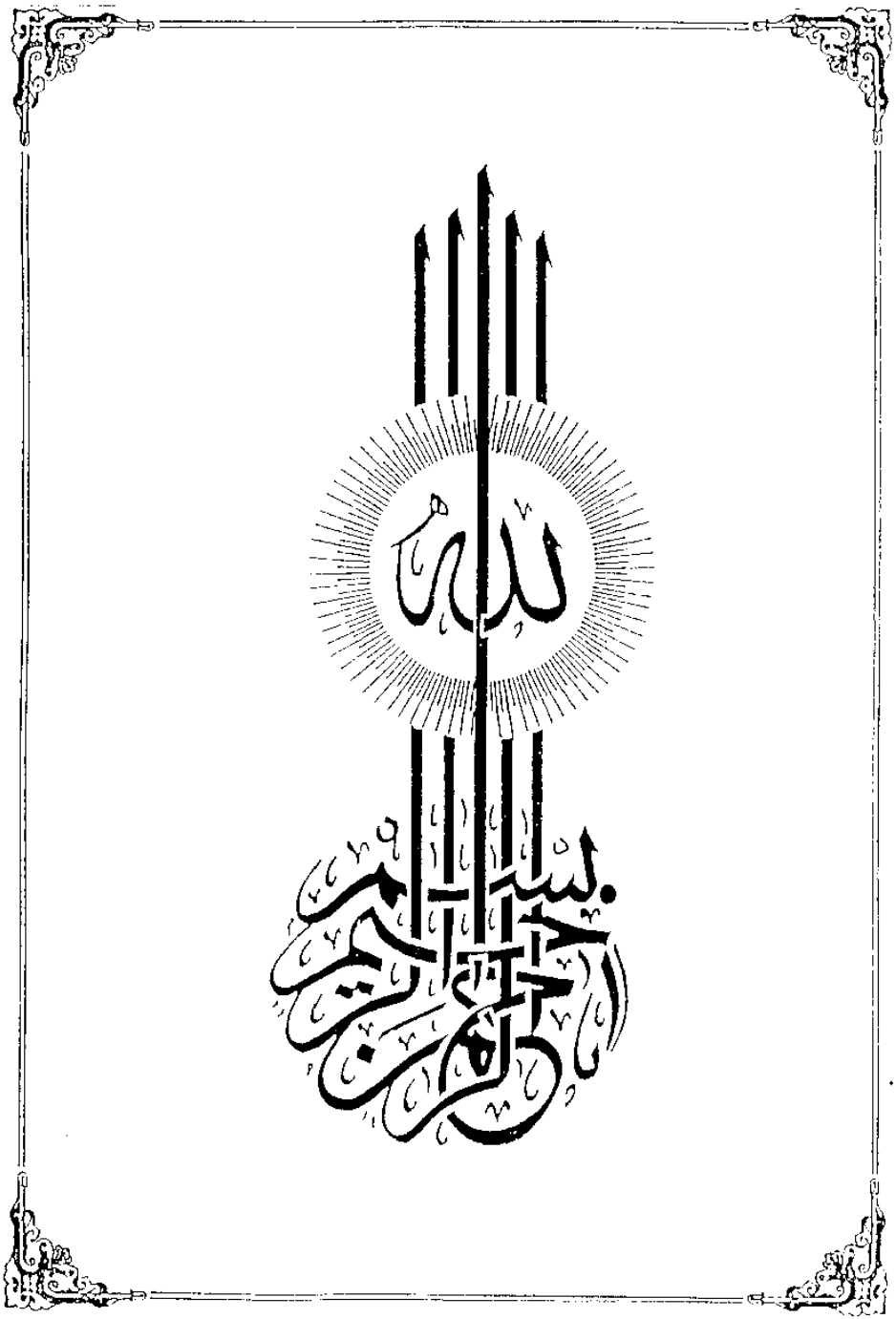

Contents

I N T R O D U C T I O N

Introduction

By nevus flammeus or port wine nevus is meant a congeni-
-tal anomaly manifesting itself as a purple discoloration of more or less extensive areas of the skin. The anatomic basis of which consists of telangiectatic vessels. This malforma-
-tion is more common on the face with the median line as its boundaries. frequently one or more branches of the trigemi-
-nal have relation to the nevus. (Mehney, 1937).

The Sturge - Weber syndrome is also described as the neurocutaneous or meningocutaneous syndrome, and is characte-
-rized by:

(1) Nevus of the skin : nevus flammeus.

(2) Angiomatous involvement of the meninges and brain.

(3) Epilepsy of Jacksonian type, usually contralateral and in some instances hemiplegia do occur.

(4) Areas of intracranial calcification.

(5) Unilateral glaucoma in many cases. (Walsh. 1957)

Etiology

Aynsley (1929) expressed the opinion that facial nevus had in itself no causal relationship with glaucoma, only when there is a general or local destruction to the branchial clefts. This produces nevi of the cerebral vessels and the mesoblastic structures of the developing eye may be affected; this may lead to imperfect forma-tion of the filtration angle or canal of Schlemm, to coloboma of the disc or choroid, or to a telangiectatic condition of parts of the intra - ocular vascular system which might sooner or later, through growth, sclerosis, transudation, or haemorrhage lead to a disturbance of balance of the intra - ocular fluids and the produc-tion of increased tension.

Again in 1929 Aynsley, Weber and Moore suggested that cases of nevi (particularly fifth nerve distribution), and cases of buph-thalmos with facial or bodily nevi, should be examined by X-rays. and examined neurologically for the presence of intracranial lesions.

A negative X-ray of the skull may simply means that the menin-geal nevus had not yet been calcified.

Aynsley (1929) favoured the opinion that the connection between facial nevus and glaucoma was accidental and an expression of the fact commonly observed that several congenital defects may be pre-sent in one person.

Werner (1952) thought that, in the Sturge - Weber syndrome as in other phacomatoses, and inherited weakness of the germinative

PATHOGENESIS AND PATHOLOGY

Pathogenesis and Pathology

The frequent ocurrence of angioma of the choroid in associa-
-tion with nevus flammeus of the eyelids and face suggests a common origin for these tumors.

Many theories have been proposed to account for the occurrence of the glaucoma in cases of nevus flammeus.

Von Barensprung (1863) was the first who reported that the nerve theory of nevus of the skin.

He stated that:

> Nevi usually appear on half the body, not overlapping the midline.
>
> They are manifested as streaks or spots on the skin which show an arrangement corresponding to the peripheral distribution of one or more spinal nerves.
>
> The degeneration of the skin depends essentially upon a hypertrophy of the skin papillae, the same structures in which the peripheral end.

Von Barensprung (1863) believed that a congenital disease of a single ganglion origination in the uterus is the cause of the skin degeneration which developed in the peripheral region of the corresponding spinal nerve.

According to Verhoeff (1903) the development of vascular nevi took place as follows:

was cupped and the tension 40 mm. It would seem more likely that a variable vascular rather than an architectural abnormality was pre--sent in this case.

Tyson (1932) suggested that the anterior chamber was blocked by a plasmoid aqucous resulting from changes in capillary permeabi--lity.

It is known that any condition which tends to dilate the intra--ocular capillaries leads to an increased protein content of the aqueous. This augmented protein content tends to produce an increased of the blood and the aqueous.

Dunply (1935) gave the proof for tyson suggestion by giving intravenous injection of glucose to one of the patient presenting with the syndrome. No effect was obtained on the intraccular tension while in a normal eye such injection would certainly have reduced the tension. This fact support the theory of abnormal permeability of intraocular capillaries.

Another proof was givin by tyson (1932) himself. He adminstered fluoresein to a petient with encephalotrigeminal angiomatosis and noted that it appeared in the aqueous of the glaucomatous eye more quickly than in the normal eye. He concluded that increased vascular permeability in the affected eye had caused increased ocular fluid density, which resulted in blockage of the iris angle by plasmoid aqueous.

It was believed that the growth of the blood vessels is regu--lated by the vasomotor nerves. During the developmental period of

the vascular system, some defect of these vasomotor nerves might impair this regulatory influence and allow an abnormal growth of blood vessels to take place, thus forming a nevus. (Dunphy, 1935).

Dunphy (1935) believed that in most cases of the glaucoma was due to an obstructed filtration angle which in turn was due to an-
-giomatous changes in the uvea or to adhesions in the iris angle resulting from the toxic action of the degenerative changes within the eyeball.

He also described a striking example of degenerative changes that occurred in one of his cases, in the form of bone formation in the choroid.

Unfortunately, Mansheim (1953) found that the glaucomatous eyes in the Sturge-Weber syndrome have normal coeffecient of outflow, so he concluded that elevated intraocular pressure was due to increased aqueous production.

Font and Ferry (1972) postulated that haemorrhage from the choroidal haemangioma may cause subretinal haemorrhage and retinal detachement with forward displacement of the iris and angle closure which is secondary to peripheral anterior synechiae caused mechani-
-cally or due to neovascularization.

More recently, Weiss (1973) found vascular hamartomas of the anterior episclera and conjunctiva in all his patients. Although, in the earlier literature, patients without episcleral angiomas were reported, these angiomas may be very subtle and occasionally diagnosed only at surgery.

Weiss (1973) hypothezised that the increased pulse pressure present on tonography was indicative of an arteriovenous fistula and a combination of elevated episcleral venous pressure, indirectly deduced by the presence of engorged vessels. A congenital angle malformation contributed in variable proportions to the elevated intraocular pressure he said.

Harley (1975) postulated that the etiology is not known, but a common primordial derivation of the meningeal, choroidal and facial vessels may explain the characteristic distribution of these con--genital lesions. The condition is rare, and complete triad of eye, skin, and intracranial lesions apparently has not been reported in more than one member of a family. Genetic transmission of the dis--order is not established.

Phelps (1978) examined 19 patients with encephalotrigeminal angiomatosis and glaucoma. All of these patients had episcleral hemangiomas, the extent of which correlated roughly with the severity of the glaucoma. Elevated episcleral venous pressure was present in 11 of 12 eyes (in which this was measured). Two patients without glaucoma had small episcleral hemangiomas. Angle deformities of a congenital nature were seen in only two patients. He concluded that an elevation in episcleral venous pressure due to arteriovenous shunts was etiologically responsible for the glaucoma. However, no predictable correlation between the episcleral and intraocular pres--sures in individual eyes was found.

Several etiologic explanation for the glaucoma have been advan--ced. Congenital malformations of the anterior chamber angle may

C L I N I C A L F E A T U R E S

Clinical features

This syndrome is also described as the neurocutaneous or menin-
-gocutaneous syndrome and it is characterized by: (1) nevus of the skin (venous angioma); (2) angiomatous involvement of the meninges and brain; (3) epilepsy of Jacksonian type, usually contralateral; in some instances hemiplegia; (4) areas of intracranial calcifica-
-tion; (5) unilateral hydrophthalmos (congenital glaucoma) in many cases. (Walsh et al, 1927)

The clinical manifestations include convulsive disorder (89), facial angioma (86), abnormal x-ray findings (63), mental retar-
-dation (54), ocular abonrmalities (37), and hemiplegia (31). (Alexander and Norman, 1960).

The presence of the vascular nevus of the face, commonly known as a "portwine stain" or facial nevus flammeus and leptomeningeal angiomas are essential to the diagnosis (Harley, 1975).

Various combinations of involved structures may occur. The trisymptomatic form classically known as Sturge-Weber syndrome af-
-fect the leptomeninges, eyes, and face. Bisymptomatic forms affect the face and eyes or face and leptomeninges. There are no descrip-
-tions of a form involving the leptomeninges and eyes and sparing the face. (Ritch, 1982).

Heredity

According to Bergstrand et al, (1936), the Sterge-Weber synd--rome was due to a congenital tendency which might be inherited, but there was not such clear evidence of heredity as in von Reck--linghausen's and von Hippel Lindau's diseases. (Luc, 1958).

Chromosomal abnormalities have been reported in some patients but most have had a normal karyotype (Hayward & Bower 1960)

The disease occurrs in all races with no sex predilection. Un--like other hamartomatous diseases, there is no evidence of genetic perdisposition or inheritance, and increased frequency of consan--giunity among parents has not been observed.(Parau et al,1961).

The influence of heredity is debatable but the evidence for genetic transmission is not firm. The fact that different members of a pedigree show various manifestations of the syndrome, some merely angiomata of the skin and other various central nervous symptoms even resulting in death in infancy in convulsions has sug--gested the possibility of an irregularly dominant gene with a very variable expression. In monozygotic twins concordant manifestations were present. There is neither sex predilection nor any preponde--rance of the occurance of the lesion on either side (Duke-Elder, 1964).