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Pathology of the Sturge-Weber Syndrome

The complete syndrome consists of an intracranial angioma, a facial angioma (nævus flammeus), and ipsilateral glaucoma, the latter usually being associated with a choroidal angioma. In 1879 William Allen Sturge (1850–1919), Physician and Pathologist at the Royal Free Hospital, described the case of a child with a right-sided angiomatous lesion involving the face, neck, thorax, lips, tongue, mouth, uvula and pharynx. The lesion had been present since birth and on examination the right eye was larger than the left, the sclerotic being vascular, the retinal veins tortuous, and the choroid darker. The onset of left-sided muscular twitchings at the age of 6 months led Sturge to suspect the presence of an intracranial lesion on the right side similar to the facial blemish.

In 1922 Frederick Parkes Weber (1863–1962), the well-known physician who during his lifetime collected a large series of rare clinical syndromes, described the case of a 22-year-old female mental defective with an extensive nævus flammeus involving the head, face, neck and trunk, being more marked on the left side. The same case was reported to the Section of Neurology of the Royal Society of Medicine in 1929 when Weber described the presence of a left-sided, calcified meningeal angioma which had been seen radiologically. In 1897 Kalischer had carried out the first post-mortem on a similar case and had found a meningeal angioma. Incomplete syndromes are much more common, e.g. nævus flammeus with glaucoma. Such a case was described in 1860 by Schirmer. Pi (1931) reviewed 31 such cases and found hydrophthalmia in 10 of them. In 1935 Dunphy described 14 cases of glaucoma or hydrophthalmia with facial angioma in which a choroidal angioma was found in 10 cases. The Sturge-Weber syndrome may be associated with ocular melanosis (Arjona 1948), and Rademacher (1961) described its association with the Klippel-Trenaunay syndrome (unilateral congenital malformation of the arteriovenous system, syndactyly, overgrowth of certain fingers, and osteohypertrophic changes of one extremity).

The metameric distribution of the Sturge-Weber syndrome is explained on a developmental basis. Streeter (1918) showed that in the development of the blood supply of the brain, the primordial vascular system splits into an inner layer supplying the brain and the retina, and into an outer layer which supplies the meninges, the choroid and the face. The common derivation of the meningeal, choroidal and facial vessels can explain malformations affecting all three vascular systems.

The facial angioma, which is present at birth in nearly every case, does not spread after making its appearance. This, of course, is a feature of congenital lesions. The fact that it follows the distribution of one or more branches of the trigeminal nerve led von Baerensprung (1863) to introduce the nervous theory of the origin of this lesion. Cushing (1906) supported it when he noted that an intracranial hæmorrhage would involve an area of the brain supplied by the same nerve. Verhoeff (1914) thought the early impairment of the influence of the vasomotor nerves on the development and growth of blood vessels might result in the formation of abnormal vascular channels. When the facial lesion involves the lid and conjunctiva, the eye is almost invariably affected. The dark red nævus flammeus is usually flat and associated with facial hypertrophy. It may involve the nasal and buccal mucosæ and the trunk, and on occasions may be bilateral, when it is sometimes associated with bilateral glaucoma. Microscopically the lesion consists of dilated vessels engorged with blood and lined by a single layer of endothelial cells.

The intracranial lesion consists of areas of calcification of the cortex which are associated with atrophy of adjacent cerebral tissue. There is a meningeal angioma consisting of racemose, dilated, hypertrophied blood vessels commonly involving the parietal and occipital lobes but which may be so extensive as to cover the whole cerebral hemisphere on one side and on occasions the other hemisphere may be affected. The underlying brain tissue shows pressure atrophy with loss of nerve cells. The lesion is congenital but may simulate neoplastic growth by rapid increase in size due to hæmorrhage. There is thickening of the overlying skull, and a similar angiomatous malformation may be present in the spinal cord.

The glaucoma may be congenital, open angle, or due to anterior synechiæ. O'Brien & Porter (1933), in a series of cases of glaucoma and facial angioma, found that the glaucoma was buphthalmic in 68% of cases. The glaucoma is usually accompanied by a choroidal angioma consisting of dilated vascular channels lined by endothelium and separated by a small amount of stroma. The congenital nature of the lesion is shown by the fact that it merges imperceptibly with the normal choroidal vasculature, it does not increase in size,

and may undergo spontaneous regression. It is usually situated between the macula and the disc; it is a flattish lesion and may be accompanied by retinal detachment, the subretinal space containing serous exudate. Choroidal detachment is frequently seen (V. d. Helm 1963, personal communication). A fibrotic membrane may be present between the tumour and the retina, and this may show pigment disturbance, calcification or ossification. The onset of glaucoma is easily explained when the retinal detachment pushes the iris forwards occluding the chamber angle. Other obvious causes include occlusion of the filtration angle by organizing hæmorrhage, occlusion of the trabecular meshwork by pigment, angle occlusion by an angioma of the iris or ciliary body, and by anterior synechiæ following anterior uveitis.

However, it may be difficult or impossible to offer an explanation when these changes are absent. Tyson (1932) thought that there was an increased capillary permeability in the ciliary body resulting in changes in the aqueous leading to occlusion of the chamber angle. This explanation has not been confirmed by experimental work. Safar (1923) and Salus (1923) thought that obstruction to the aqueous outflow was due to abnormal tissue in the chamber angle similar to that found in congenital glaucoma. Barkan (1954) described 2 cases of nævus flammeus associated with glaucoma in which there was occlusion of the filtration angle by abnormal tissue suggesting a mesodermal type of chamber angle as seen in congenital glaucoma. He was able to reduce the intraocular tension to normal limits in both cases by dissecting this abnormal tissue in the angle at repeated goniotomies. In adults, when the glaucoma may be late in appearing, it is possible that there may be compression of one or more of the vortex veins by a choroidal angioma, or by interference of choroidal drainage by a coexisting orbital angioma, or else by interference of the drainage from the cavernous sinus by a meningeal angioma. There may be interference with drainage of the aqueous by dilated conjunctival vessels or by a hæmangioma of the conjunctiva and episclera.

Other lesions sometimes seen are cataract, corneal degeneration, uveitis, intraocular hæmorrhage, and heterochromia of the iris.

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(1929) *Proc. R. Soc. Med.* **22**, 431

Mr Murray Falconer (*Maudsley Hospital, London*) read a paper on the **Neurosurgical Aspects of the Sturge-Weber Syndrome.** The material he presented has been covered by the following papers:

Falconer M A (1961) In: Hemiplegic Cerebral Palsy in Children and Adults. Little Club Clinics in Developmental Medicine No. 4. National Spastics Society, London; p 140
Falconer M A & Rushworth R G (1960) *Arch. Dis. Childh.* **35**, 433

Meeting October 11 1962

The following papers were read:

A Note on the Troncoso Method of Gonioscopy
Mr John Foster (*Leeds*)

Presbyopia: New Light on Old Eyes
Dr R A Weale (*London*)

Recent Advances in Anterior Chamber Implant Technology, Illustrated by Cases Shown
Mr D P Choyce (*London & Southend*)
(*see Lancet*, 1963, i, 794)

Meeting November 8 1962

The following conditions were demonstrated and discussed:

Schilder's Disease
Dr J M L Howat and Dr R Barry

Chondrodystrophia Calcificans Congenita
Dr J Luder and Dr J Harry

Ocular Ochronosis
Dr F D McAuley and Professor Norman Ashton

An Electron Microscopic Study of Pseudo-exfoliation of the Lens Capsule
Dr R T Collyer

Meeting December 13 1962

Surgical Minutiæ

The following short papers were read:

(1) **A Method of Repair of a Torn Canaliculus**
(2) **Procedure for Alleviating Lacrimal Obstruction**
Mr Peter Wilson (*Huddersfield*)

Sutures
Mr Redmond Smith (*London*)

Technique in Trephining
Mr F W Law (*London*)

The Use of the Head Lamp in Ophthalmic Surgery
Mr R A Burn (*London*)

Splitting the Lid in the Kuhnt-Szymanowski Operation
Mr J D Abrams (*London*)

A Simple Keratome and Scissors Subconjunctival Incision for Total Cataract Extraction
Mr J E H Cogan

Some Prototype Instruments for Cataract Surgery
Mr Dermot Pierse (*London*)

Retinal Drainage
Mr M Gilkes

The Technique of Drainage of the Subretinal Fluid
Mr Lorimer Fison (*London*)

A Method of Cataract Extraction Using Alphachymotrypsin with a Small Pupil
Mr D P Choyce (*London & Southend*)

A New Ptosis Knife
Sir Benjamin Rycroft (*London*)

The use of a new ptosis knife for the correction of ptosis by an autograft was illustrated: the Werb right-angle scissors for dacryocystorhinostomy and special polythene tubes for canalicular obstruction were also shown. Speaking on corneal graft surgery, Sir Benjamin stressed the importance of deep suture fixation to ensure apposition of the endothelium in penetration grafts, and also pointed out that the fixation of a splint by overlay sutures tended to close the host graft junction more effectively than fixation of the splint with direct sutures.

Meeting January 10 1963

A Forum was arranged in which the following took part: Sir Benjamin Rycroft, Mr Willoughby Cashell, Mr Nigel Cridland, Professor W J B Riddell and Mr Kenneth Wybar.

Meeting March 22 1963
A joint clinical meeting of the Section with the Southern Ophthalmological Society was held at the Sussex Eye Hospital, Brighton. Cases were shown and discussed.