assay has helped resolve this problem. C-peptide and insulin levels are measured when the patient is hypoglycaemic. Because endogenously secreted insulin is accompanied by the liberation of C peptide, while exogenous insulin will suppress endogenous secretion and hence lower the C-peptide concentration, serum insulin and C-peptide assays will show similar elevations in pancreatic hyperinsulinism, while C-peptide levels will be low in relation to the serum insulin level after injection of exogenous insulin.\textsuperscript{4}

The patient has been discharged from hospital on insulin therapy and has been requested to attend the diabetic clinic in future. Our hope is that he will not persist with his deception as do so many patients with Munchausen’s syndrome.

REFERENCES


Encephalo-angiomatosis in Black children
A report of 2 cases

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Summary

Two cases of encephalo-angiomatosis are reported. The clinical presentation and certain neurological findings and investigations confirm the diagnosis of a disease which is relatively rare among Blacks.

The case reports were considered valuable because of the rareness of the condition. Up to 1962 only 145 cases had been reported in the world literature. Encephalo-angiomatosis or Sturge-Weber syndrome is one of the disease groups known as phakomatoses, and an attempt is made to characterize each phakomatosis condition.

Encephalo-angiomatosis or Sturge-Weber syndrome belongs to the group of diseases that falls under the umbrella term of phakomatosis. Phakomatoses are defined as genetically determined neuro-ectodermal lesions of the central nervous system. Among the phakomatoses are neurofibromatosis, tuberous sclerosis, neuroretinal angiomatosis (von Hippel-Landau disease), neurocutaneous melanosis, Sturge-Kalischer-Weber disease, Klippel-Trenaunay-Weber syndrome, and encephalo-angiomatosis. Phakomatosis (Greek for 'lentil-shaped spots') was the term coined for certain heredofamilial tumorous states characterized by lens-like masses.

Neurofibromatosis and tuberous sclerosis are mendelian dominant conditions and are the most common forms of phakomatosis. Von Hippel-Lindau disease is a rare affliction but where it occurs in a family, very few members escape the 'mother's spot'.

It is most unlikely that these conditions will be confused with encephalo-angiomatosis or Sturge-Weber syndrome, since there are certain distinct characteristics such as the 'cafe au lait' spots, the molluscum fibrosum and sarcomatous degeneration of skin, subcutaneous tissue and peripheral nerves in neurofibromatosis. The glial nodules in brain and retina and the almost invariable accompaniment of adenoma sebaceum in a mentally retarded child make the diagnosis of tuberous sclerosis obvious. The neuroretinal angiomatosis with the 'trigeminal' port-wine stain, and the almost invariably subpentental haemangiblastoma in a member of a family with a strong familial trait, make the diagnosis of von Hippel-Lindau disease obvious.

Sturge-Weber syndrome

The first description of this condition was given in 1879 by Sturge, followed by Kalischer in 1887, who also reported the autopsy he performed in 1901. Parkes Weber added to the literature in 1922 the lucid description of the condition bearing the name Sturge-Weber syndrome.

Sturge-Weber syndrome is characterized by cutaneous capillary malformation of the face and atrophy of the ipsilateral hemisphere with gyral calcification in a subcortical telangiectatic malformation and contralateral underdevelopment of limbs. Buphthalmos, hemiparesis and epilepsy are encountered.\textsuperscript{1} The buphthalmos is of necessity a condition that develops during intra-uterine life or shortly after birth on account of the resistance offered by the mature scleral fibres, preventing enlargement of the eye.

The telangiectases in the subcortical layers have a tendency to calcify in the characteristic 'tramline' fashion, following the gyri and giving a diffuse, wavy appearance outlining the atrophic hemisphere on plain radiographs of the skull. In longstanding cortical atrophy it is associated with reduced size of the hemicranium (the so-called Dyke-Davidoff syndrome), which distinguishes it from Sturge-Kalischer disease.

Angiography demonstrates angiomas and telangiectasia. The syndrome is unlike an arteriovenous anomaly except that the absence of draining veins is a characteristic feature of both; it
demonstrates a defect of the blood-brain barrier with reduced circulation which is responsible for the atrophy of the brain and the absence of draining veins.

The computed tomography (CT) scan demonstrates the cerebral abnormalities more accurately and with greater sensitivity — the gyriform calcification, ventricular enlargement and cortical atrophy. Enhancement causes a diffuse homogeneous haze of the cortex.

Sturge-Weber syndrome is usually unilateral, but cases of bilateral intracranial calcification have been reported. It is predominantly a childhood disease which manifests at between 3 and 10 years of age, although the abnormalities will have been present from birth. The skin telangiectasia also grows in size with age, and it is usually late in childhood that underdevelopment of the contralateral limbs is noticeable.

Vouge et al. reported a case of Sturge-Weber syndrome associated with tuberous sclerosis. The condition usually manifests in early childhood, but intra-uterine calcification has been reported in a neonate.

Differential diagnosis

The following conditions, the first two of which are phakomatoses, may cause some confusion, but in their pure form have identifiable characteristics:

**Sturge-Kalischer-Weber disease** is a curious haemangiectatic condition involving the face and meninges. The haemangiomatous malformation is confined to the meninges and the face, and can therefore be differentiated from von Hippel-Lindau disease where the malformation is situated in the brain substance and also from Sturge-Weber syndrome which has a characteristic subcortical 'tramline' calcification and diffuse telangiectatic subcortical abnormality.

**Klippel-Trenaunay-Weber syndrome** is characterized by arteriovenous malformation of the cerebral hemisphere associated with hypertrophy of the hemisphere. The contralateral limb is usually involved, characterized by cutaneous angiomatous malformation and hypertrophy of the limb.

**Meningioma** may be associated with disseminated plaques of arachnoidal calcification.

Gyriform calcification after purulent meningitis has been reported, but the calcinosis was bifrontal instead of the unilateral parietal calcinosis in Sturge-Weber syndrome.

**Congenital toxoplasmosis** can give rise to intracranial calcification, chorioretinitis, mental retardation and epilepsy. Disseminated encephalomyelitis associated with necrotic softening of the meninges leads to flake-like subcortical calcification. The skin and serological tests should differentiate between this condition and Sturge-Weber syndrome.

**Systemic lupus erythematosus** associated with intracranial calcinosis is unlikely to be confused with Sturge-Weber syndrome, on account of the skin manifestations.

**Calcification of a subdural haematomata** usually occurs in the old and is therefore unlikely to be confused with Sturge-Weber syndrome of childhood.

**Atypical forms of phakomatosis** have been reported where typical subependymal calcification as seen in tuberous sclerosis and subcortical calcification as seen in Sturge-Weber syndrome are present.

**The coexistence of a cerebral tumour** and Sturge-Weber syndrome has been reported.

The intracranial calcinosis seen in patients with leukaemia who received intrathecal methotrexate and irradiation simulates the intracranial calcification seen in Sturge-Weber syndrome.

The same authors reported a patient with coeliac disease and epilepsy treated with anticonvulsants who developed cortical calcification identical to that in Sturge-Weber syndrome. They postulated that the calcification may have been secondary to folic acid deficiency interfering with metabolism in the central nervous system.

Gyriform calcification has been described following an attack of encephalitis.

Case reports

Case 1

A 15-year-old Black boy was admitted to Ga-Rankuwa Hospital on 5 November 1981, his second admission within 6 months.

At his first admission post-epileptic stupor was diagnosed and he was discharged after 1 week on anticonvulsant therapy. Subsequently it was ascertained that he had taken a large dose of phenobarbitone tablets before his admission to hospital.

After his second admission he was stuporous for 4 days, and once again a history of overconsumption of tablets was given. Since he had mental retardation, we decided that both overdoses were not suicide attempts but that the ingestion was accidental during periods of confusion. After his confusion had cleared up, his condition was studied more fully.

The following history was given by his grandmother: Birth was normal. During the first 3 months of life it was noticed that he often lay with his head turned to the left side. The grandmother would then gradually straighten it over a period of several days.

Epileptiform seizures started after the 3rd month, up to four attacks in 24 hours. These were controlled by medication. Soon after birth the right leg and arm were noted to be abnormally weak. She also noted discolouration of the left side of the face in the neonatal period, over the forehead, nose, cheek and both lips. This abnormality stopped at the midline. Mental retardation was apparent during the first few years and has remained constant. The seizures had worsened during the last year.

A large flat haemangioma was present on the left side over the forehead, cheek, nose and both lips; this stopped abruptly at the midline. It was present inside the lips, and on the left side of the palate (Fig. 1), also with midline demarcation. The lesion on the skin was difficult to see unless studied closely.

**Fig. 1.** The telangiectatic discolouration (arrowed) with a midline demarcation is clearly seen.

There was left-sided peri-orbital swelling, with buphthalmos and conjunctival involvement. The fundus was grossly abnormal, but there was no glaucoma (Fig. 2). Skull radiographs and angiography revealed the typical calcified festooned appearance of the syndrome (Fig. 3). There was also an old healed fracture of the left clavicle. An apical root abscess of the lower jaw on the right side accounted for pain and swelling at that site. The tooth was removed.
Fig. 2. Funduscopy shows marked glial patches involving the disc margins and spreading to the periphery.

Fig. 3. Angiogram (lateral projection) showing gyral calcification and diffuse cortical uptake of the contrast material. There is also evidence of cortical atrophy.

Fig. 4. Underdevelopment of the right arm and leg. The right leg is at least 3 cm shorter than the left leg.

He was unable to use his right hand owing to a severe flexion contracture of the right wrist. There was hemi-atrophy of the right upper and lower limbs with paresis of the extremities (Fig. 4). The leg and thigh disability caused a limp on the right side. All reflexes were exaggerated on the right side and marked ankle clonus was present. The plantar reflex was equivocal.

Psychiatric consultation confirmed the mental retardation, and his growth and development were retarded for his age. The diagnosis of Sturge-Weber syndrome, with characteristic stigmata of the condition, was made. There were further seizures while he was in hospital and therapy with an increased dosage of medication was commenced.

Case 2

A 4-year-old girl was referred with a history of swelling of the left side of the face since birth. She had had a normal birth and normal milestones and there was no history of seizures.

On examination she was found to be slightly mentally retarded. The left side of the face was hypertrophied with increased vascularity, and was warmer and darker in colour. The left arm was longer and thicker. The thenar eminence was bright red in colour because of a small telangiectatic lesion. The disparity between the limbs was due to underdevelopment of the limbs on the right side and not due to hypertrophy of the limbs on the left side.

Full blood count and urinalysis revealed no abnormality. On skull radiographs calcification in the cortex was just visible. Underdevelopment of the left half of the cranium was noted, similar to that in case 1. Angiography revealed a blush diagnostic of Sturge-Weber syndrome (Figs 5 and 6). A CT scan revealed typical calcification of gyri, with associated hemispherical atrophy and underdevelopment of the left side of the cranium (Figs 7 and 8).

Discussion

Encephalo-angiomatosis or Sturge-Weber syndrome is one of a group of conditions referred to as phakomatoses, genetically determined neuro-ectodermal lesions of the central nervous system. Each entity as originally described has certain characteristics, but the coexistence of two or more entities has been reported. An attempt is made to delineate the characteristics of each syndrome clearly so that no confusion can exist as to the category to which a case in question should be correctly allocated.

Case 1 leaves one with no doubt as to the correct classification. In case 2, although all features are consistent with the diagnosis of Sturge-Weber syndrome, the telangiectatic patch on the left thenar eminence causes one to consider the possibility of Klippel-Trenaunay-Weber syndrome, but a firm diagnosis of Sturge-Weber syndrome was made in the light of the underdevelopment of the skull on the left side, the atrophy of the left hemisphere and underdevelopment of the contralateral limbs.

Table I shows the characteristics for each of the conditions falling under the umbrella term of phakomatosis, with the exception of neurocutaneous melanosis which is extremely rare and easy to diagnose.
<table>
<thead>
<tr>
<th>Condition</th>
<th>Genetically determined ectodermal lesion</th>
<th>Symptoms</th>
<th>Signs</th>
<th>Skull/spine radiographs</th>
<th>Angiograms</th>
<th>CT scan</th>
<th>Definitive diagnosis</th>
</tr>
</thead>
<tbody>
<tr>
<td>Neurofibromatosis (von Recklinghausen's disease)</td>
<td>+</td>
<td>Skin lesions, scoliosis, paresis, deafness, etc.</td>
<td>Café au lait spots, molluscum fibrosum</td>
<td>No characteristics of skull but enlargement of intervertebral foramen</td>
<td>Nonspecific, except skin and retinal lesions</td>
<td>'Candle-wax' sign on ventriculography</td>
<td>Tuberous lesions projecting into ventricles</td>
</tr>
<tr>
<td>Tuberous sclerosis (Bourneville's disease)</td>
<td>+</td>
<td>Adenoma sebaceum of face</td>
<td>Epilepsy and mental retardation</td>
<td>Nonspecific</td>
<td>Capillary angiomatosism of the cerebellum</td>
<td>May demonstrate the arteriovenous anomaly</td>
<td>Cutaneous cerebellar signs</td>
</tr>
<tr>
<td>Neuroretinal angiomatosis (von Hippel-Lindau disease)</td>
<td>++</td>
<td>Port-wine stain on face, epilepsy, cerebellar signs, subarachnoid haemorrhage</td>
<td>From port-wine stain on the face to blood in CSF</td>
<td>Nonspecific</td>
<td>Linear calcification of the dura mater</td>
<td>Nonspecific</td>
<td>Calcification of the dura mater</td>
</tr>
<tr>
<td>Sturge-Kalischer-Weber syndrome</td>
<td>+</td>
<td>Mental retardation, fits</td>
<td>Telangiectasia of face or limbs</td>
<td>Hypertrophy of ipsilateral hemicranium</td>
<td>Hypertrrophy of hemisphere, angiomatosism, malformation of brain and contra-lateral limbs</td>
<td>Calcification of the dura mater</td>
<td>Skull enlarged on the ipsilateral side and contralateral limb hypertrophy</td>
</tr>
<tr>
<td>Klippel-Trenaunay-Weber syndrome</td>
<td>+</td>
<td>Fits and mental retardation</td>
<td>Telangiectasia of face afd limbs, overdevelopment of contralateral limbs</td>
<td>Hypertrrophy of hemisphere, angiomatosism, malformation of brain and contra-lateral limbs</td>
<td>Hemispherical atrophy associated with ipsilateral underdevelopment of cranium, gyral calcification and diffuse cortical enhancement</td>
<td>Underdevelopment of cranium on ipsilateral side, associated with ipsilateral cerebral atrophy and contra-lateral underdevelopment of limbs</td>
<td></td>
</tr>
<tr>
<td>Encephalo-angiomatosis (Sturge-Weber syndrome)</td>
<td>+</td>
<td>Telangiectasia of face, buphthalmos, fits and hemiparesis</td>
<td>Port-wine stain of skull and contra-lateral hemiparesis</td>
<td>Underdevelopment of skull and contra-lateral development of limbs, calcification of gyri</td>
<td>Hemispherical atrophy and diffuse staining of cortex and absence of drainage veins</td>
<td>Underdevelopment of cranium on ipsilateral side, associated with ipsilateral cerebral atrophy and contra-lateral underdevelopment of limbs</td>
<td>Underdevelopment of cranium on ipsilateral side, associated with ipsilateral cerebral atrophy and contra-lateral underdevelopment of limbs</td>
</tr>
</tbody>
</table>
Conclusion

The clinical picture of Sturge-Weber syndrome is distinct from Klippel-Trenaunay-Weber syndrome, Sturge-Kalischer-Weber syndrome or tuberous sclerosis. No difficulty should be encountered in making a firm diagnosis.

REFERENCES