

Background: Sturge–Weber Syndrome (SWS) is a neurocutaneous syndrome, often diagnosed in childhood, defined by the association of a facial capillary malformation (port-wine stain) with vascular malformation of the eye, and/or vascular malformation of the brain (leptomeningeal angioma). This definition is referred to the complete form, however there are two more types of SWS: type 2, involving a facial angioma and the possibility of glaucoma, but no evidence of intracranial disease, and type 3, characterized by leptomeningeal angioma, with no cutaneous and/or ocular involvement.

Case report: We report the case of a 56 years male with a history unremarkable except for a few episodes of loss of contact during the last 2 months, who was admitted to our hospital because of a secondary generalized seizure, starting from right arm. Neurological and physical examination, blood tests and electroencephalography (EEG) revealed no abnormalities. The brain magnetic resonance imaging (MRI) put in evidence, after gadolinium injection, a pial enhancement along cortical sulci of left cerebral hemisphere, consistent with a leptomeningeal angioma. In the same locations, the head computed tomography (CT) pointed out gyriform calcifications. Imaging data, associated with epileptic manifestations, allowed us to make diagnosis of SWS type 3. We started anticonvulsivant therapy with phenobarbital 125 mg/die and prophylactic antiaggregant therapy. At the moment, 1 year after, the patient is still free from seizures.

Conclusions: There are no studies to date reporting the case of a patient who receives his first diagnosis of SWS in the sixth decade. Some authors hypothesized that presentation of SWS variants at an older age than classical form was due to a less extensive insult to vascular development, that would result in milder impairment in brain development and function. We can assume that, in our case, involvement of the meninges only, with sparing of skin and eye, is indicative of a less extensive vascular insult, and has allowed the absence of seizures and normal development of the brain until such an advanced age.

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Seizures and tics with a similar motor phenomenology

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Introduction: Tics are sudden, fast, recurrent, arrhythmic, stereotyped, irresistible motor movements or vocalizations, partly dependent on one's will. They can be primary or secondary. Tics have been described after epilepsy surgery, but not in patients with symptomatic epilepsy sharing characteristics of ictal semiology.

Case-report: A 40-year-old man with a past history of traumatic brain injury in childhood and epileptic seizures since 5. The seizures begin with a smell "as in the hospital", followed by loss of consciousness, right head rotation, oroalimentary and manual automatic movements, predominantly on the left side, lasting 30–60 s followed by post ictal confusion, hunger and amnesia for the event. The seizures were not controlled under treatment with Levetiracetam 1,000 mg/day and Sodium Valproate 2,000 mg/day. He also displays stereotyped movements of lateral contraction of the nose and bilateral blinking, partly controlled by will and exacerbated with hyperpnia, followed by a sense of relief, and arising particularly in situations of distractibility. MRI shows right mesial temporal sclerosis and a small, nodular, neocortical, lesion in the right parietal convexity. Video-EEG shows seizures beginning with blinking and lateral contraction of the nose, similar to the previously described tics, followed by manual and oroalimentary automatism and predominantly right cephalic version. Ictal scalp EEG changes begin in the fronto-temporal and right inferior temporal regions, 15–30 s after clinical onset. Back-average analysis didn't show any cortical correlate for the tics.

Discussion: We present a case of symptomatic epilepsy associated with simple motor tics, in which seizures and tics have a similar phenomenology. The analogy between ictal motor patterns and parasomnias has been associated with activation of central motor generators by different mechanisms. This case suggests that tics may also share motor patterns with seizures, probably related to subcortical activation. Future studies should assess the frequency of tics in patients with epilepsy.

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An estimation of the 2010 incidence of hyperammonaemic encephalopathy related to endovenous valproate seen at a level II Hospital in Catalonia, Spain

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Objectives: ev VPA is an antiepileptic drug of common use. HE is a well known adverse event (AE) of VPA, especially when used with old age patients, known liver disease, polytherapy and endovenously. Our target is to estimate the incidence of this AE in our daily practice.

Methods: We retrospectively reviewed the clinical files of all the in-patients who had been under treatment with ev VPA throughout 2010 (January to December) at our hospital; a level II (150 beds and with specialist in formation occasionally) hospital of the Spanish Public Health System.

Results: During 2010, 33 in-patients (18 male/14 female, median age (MA) 68.6 years, 93% under polytherapy) received ev VPA. VPA was started for the first time in 26 patients. Of those; 3 patients (11%, 2 male/1 female, MA 75 years) developed an HE. The maximum ammonium levels were 105, 70, 90 $\mu\text{mol/L}$ (n: 9–33). All of them received other drugs. 2 did have known liver disease and showed hyperammonemia without impairment of other markers of liver function (AST, ALT, GGT, FA). 1 patient did not have known liver disease and showed significant impairment of markers of liver function. All patients did normalize their ammonium levels when VPA was retired. All patients improved substantially their level of consciousness (1 complete recovering) when VPA was discontinued.

Conclusions: Our estimate annual incidence of HE has been relatively high (11%). We think VPA is an excellent first line antiepileptic drug, but we have to be careful when using ev VPA, specially when dealing with patients of old age, under polytherapy, and known liver disease.

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Progressive myoclonic epilepsy: searching for a genetic diagnosis

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Introduction: Progressive Myoclonic Epilepsies (PME) are a group of diseases of which the majority have a genetic cause, with variable age of onset, classically characterized by myoclonic seizures, tonic-clonic seizures, ataxia and cognitive decline. Although myoclonus is the predominant manifestation, age of onset and association with other neurological and non-neurological symptoms and signs help to guide aetiological investigation.

Methods and results: We describe two siblings (absent consanguinity in the family): a 29-year-old man and a 31-year-old woman with multifocal myoclonus which began at age 16 and 19,

respectively, present during action and posture, sensitive to light, with progressive worsening, refractory to anticonvulsants, leading to inability to walk unaided 5 years later. During follow-up a pancerebellar syndrome gradually developed, action dystonia of distal upper limbs was observed in the woman, the man developed generalized tonic-clonic seizures and no cognitive decline or ophthalmological abnormalities were present. Video-EEG monitoring revealed a normal background activity, myoclonus without EEG correlation and generalized myoclonic seizures with epileptiform discharges. Brain MRI was normal. Enzyme studies were normal (*N*-acetyl neuraminidase, β -galactosialidase), as were urinary oligosaccharides. Skin and muscle pathology were normal (including electronic microscopy, periodic-acid-Schiff staining, and absent red ragged fibers). Genetic study for MERRF mutations and CAG expansion in DRPLA gene were negative. Finally, CSTB repeat expansion was also negative.

Discussion: Pedigree suggests an autosomal-recessive pattern of inheritance and phenotype is similar to the one observed in Unverricht-Lundborg Disease (ULD), even though investigation was negative for CSTB repeat expansion and other PME causes. While a homozygous mutation expansion is found in about 90% of patients with ULD, its absence does not exclude the diagnosis, since a recent study provides evidence for genetic heterogeneity in patients with ULD phenotype.

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Epileptic seizures precursor of cerebral haematomas

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Objective: To demonstrate that the relationship between epilepsy and cerebrovascular disease is not restricted to post-stroke epilepsy, but also involves the concept of “vascular precursor epilepsy”. These heralding seizures are usually related with ischemic stroke but a few cases of primary cerebral hemorrhage have been reported in the literature.

Method: Case report and literature review.

Results: We report a case of 82-year-old female, without previous neurological complaints, who presented transient episodes (lasting <24 h) of left hemiparesis, homonymous hemianopsia and inattention to the hemi-space. Afterwards, she evolved to paroxysmic episodes of clonic and dystonic position of the left arm, sometimes with secondary generalization, and post-critical left hemiparesis and inattention to the hemi-space. These episodes led to the introduction of anti-epileptic therapy. She also had two episodes of prolonged confusional status, which responded to anti-epileptic regime modification, regarded as non-convulsive status epilepticus. EEGs showed a right hemispheric dysfunction and focal slow activity with very frequent epileptiform discharges in right temporal, posterior temporal, parietal and occipital leads. Several cranial CT scans and CSF were unremarkable. A 0.5T brain MRI was performed, as the patient had a metal prosthesis, but it was not conclusive because she was not cooperative. Two and a half years later, a new episode of prostration and a left hemiparesis led to a new cranial CT scan, which showed a right anterior frontal cortico-subcortical haematoma. Traumatic and hypertensive cause were excluded and CT angiography did not show cerebral aneurysms, arteriovenous malformations and abnormal venous drainage. The EEG revealed bilateral independent epileptiform discharges on right temporal and left fronto-temporal regions. Two months later, she was admitted with seizures and persistent right hemiparesis and an extensive left temporo-parietal haematoma was diagnosed. The EEG disclosed bilateral independent epileptiform activity on right frontal, temporal and posterior temporal and left frontal regions. The patient died 1 month later.

Conclusion: This case exemplifies the concept of “vascular precursor epilepsy”. The investigation suggested the diagnosis of probable cerebral amyloid angiopathy as a cause of recurrent lobar hemorrhages. This support the hypothesis that pathological changes are also related to the previous epileptic events.

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Epilepsy in focal cortical dysplasia—clinical characterisation

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Objective: Malformations of cortical development (MCD) are a heterogeneous group characterized by structural abnormalities of the cerebral cortex. Within this group we have Focal Cortical Dysplasia (FCD), an entity that results from an error during cortical cell proliferation and differentiation. First recognized in 1971 as an epileptogenic lesion, FCD has been recognised as a major cause of refractory epilepsy, especially in children. Our aim is to characterize the group of patients with epilepsy and FCD followed in our neurology department.

Methods: We reviewed the Brain MRI and Clinical Files of all patients followed in our outpatient clinic and Epilepsy Monitoring Unit. Then we selected those who had an image consistent with FCD.

Results: We found 45 patients with MCD and 32 (71%) with FCD. Of these, 23 were women (71.8%) with an average age of 33.45 years old. The average age of epilepsy onset was 12.7 years. The majority (55%) had at least weekly seizures and 34% had daily seizures. About 80% of the patients were receiving polytherapy, with 62% taking at least three antiepileptic drugs. Two patients had dual pathology, more precisely mesial temporal sclerosis associated with ipsilateral temporal dysplasia. Most patients (84%) had an extra-temporal localization.

Conclusion: As already noted by many other authors, epilepsy associated with FCD is usually refractory. It's interesting to note that as in other reviews, we found that when dysplasia is associated with mesial sclerosis, it tends to be located in the temporal lobe. It's important to remember the bias inherent to the limitations of current diagnostic methods regarding the detection of dysplasia in more innocent histological patterns, which will correlate with more discrete imaging abnormalities and a potentially more benign clinical presentation.

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Amniotic fluid embolism: report of a case presenting as status epilepticus

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Introduction: Amniotic fluid embolism is one of the most catastrophic complications of pregnancy. Although its rarity, it is one of the differential diagnosis of seizures occurring during labor.

Case report: A healthy, 35 years old, 40 weeks pregnant woman developed generalized tonic-clonic status epilepticus, during the labor, evolving to bradycardia and respiratory arrest. An emergent cesarean section was performed resulting in a healthy newborn. After surgery patient was found to be in cardiogenic shock (peripartum cardiomyopathy confirmed by ultrasound) and multiple organ dysfunction, namely respiratory, hepatic, hematological (without thrombocytopenia or coagulation abnormalities), and metabolic.