Index

Abstract **1**

Introduction **3**

Case report **4**

-History 4

-Evaluation and Pathological findings 5

-Surgical treatment 5

-Postoperative course 5

Discussion **6**

Acknowledgments **7**

Disclosure **7**

References **7**

Figures **9**

Appendix 1 ..... **10**

Case-report: Portuguese version

Appendix 2 ..... **14**

Poster form that were presented in the National Annual Congress of Portuguese Society of Neurosurgery

Keywords: *Epilepsy, Disconnective surgery, refractory seizures, Sturge-Weber Syndrome*

Abstract

Sturge-Weber Syndrome (SWS) is a rare neurocutaneous syndrome characterized by vascular malformations involving skin, brain and sometimes the eyes. These patients usually have venous angiomatosis of the leptomeninges and ipsilateral facial angiomatosis appearing very frequently as a port-wine nevus of the skin[1.] Seizures are one of the main features of SWS patients, sometimes refractory, occurring in a high percentage but the control achieved with the antiepileptic drugs is a little bit disappointing. Surgery must be considered in patients with SWS refractory epilepsy with the aim of seizure control.[2] Many surgical options are described to treat SWS patients, the classic types being lobectomy or functional hemispherectomy. We report a successful case of disconnective surgery in 24 years-old woman with an intractable epilepsy caused by SWS. The outcome was excellent, which make us believe that this procedure must be considered in other selected patients.

Introduction

Being described as a congenital disease, Sturge-Weber syndrome (SWS) is characterized by a facial capillary-venous malformation (port wine stain) with the involvement of the brain (leptomeningeal angioma) and eye[3]. These vascular malformations are associated with specific neurologic and ocular abnormalities. Neurologic features may be progressive and include seizures with some degree of focal neurological deficit, typically hemiparesis or new onset visual field deficit, when occipital cortex is affected (lasting several days, weeks or months), designated as a stroke-like episode[4].

Seizures occur up to 85%, and less of 50% are controlled under antiepileptic drugs. In addition roughly 75% of SWS patients have unilateral brain involvement and in approximately 95% of patients with the brain involvement is bilateral[5].

Hypoplastic cortical vessels related to enlarged, tortuous leptomeningeal vessels and, frequently, with dilated deep venous drainage consist in the vascular malformation of the brain[1]. Not only trigger seizures, in which chronic anticonvulsant therapy is crucial (although is not effective in many patients) but also represent a major surgical problem because of the highest bleeding risk associated with an increase in morbidity and mortality as well.

Even many series demonstrating that surgical resection is effective, and although early hemispherectomy has been recommended, there is some kind of controversy about which patients should be selected and the timing of surgery.[1]

The mainstay of surgery with strict medical indication are refractory seizures.

For the long-term seizure freedom, complete resection of the involved brain regions was requested, but this approach may increase the risk of morbidity and a lot of side effects like visual field deficit and glaucoma.

Having a better understanding about this syndrome and knowing new tools are being developed to aid either in diagnosis or in treatment in order to promote a better quality of life and a better prognosis. For this reason, we present the advantage of using disconnective technique with lower surgical risks becoming a different approach in the treatment of these patients with refractory seizures with very good results at short-term as well as long-term.

Case Report

History

24 years-old woman, dextra, with the eighth grade that has refractory epilepsy due to SWS followed in our outpatient clinic since 1988. She has alopecia in right parietal region. She has not port-wine angiomatosis skin lesions.

Seizures began when she was 2 years-old, first seizure occurred in a febrile context with clonias on the left lower limb with partial loss of consciousness. In the following days she had hemiclonics seizures of the left limbs which lasting about 20 minutes without loss of consciousness.

She referred sometimes a vertiginous sensation before seizures as well as she saw all dark. After that she had manual automatisms and clonic movements of left limbs and impairment of consciousness. She had a post-ictal left hemiparesis.

Despite different antiepileptic regimens tried she had daily and very disabling seizures with lots of falls and severe injuries during all this years, so a surgical option was considered.

Evaluation and Pathological findings

During the patient follow-up, three magnetic resonances (MRI) were performed in 1996, 2008 and 2009. The latter was found to be superimposed on previous injuries. All those images showed two lesions with different sizes. The biggest one had an extensive calcification at right medial temporo-occipital area and atrophy/gliosis of subjacent parenchyma; the second one is another calcified lesion located in right temporal convexity (figure 1).

Other exams were performed in 2007 like inter-critical EEGs and SPECTs (Inter-Ictal and Ictal). In the former several tip and sharp-wave/slow-wave located at frontal area were shown with frequent lateralization in both sides of the brain with generalization as well; in both SPECTs were observed an entire hipoperfusion predominating in the temporo-parietal-occipital area on the right side and also in the frontal left side of the brain. The hiperdebits were seen on the right side of the brain chiefly in the temporal and then in the parietal and frontal regions (figure 2A).

Numerous outbreaks of right frontal tip-wave were registered in monitoring video-EEG carried out in 2008: she felt a general malaise and a vertigo losing her consciousness soon after. Left limbs were used fewer than right ones, showed as well by functional

MRI albeit we couldn´t have the perception if it was an hemineglect or a hemiparesis (figure.2B and 2C). She had a left post-ictal hemiparesis.
Humphrey visual field assessment showed a lower left homonymous quadrantanopia.

Surgical treatment

Patient underwent on surgery on September 2010. Craniotomy was performed and right temporo-parieto-occipital region was exposed. The extensive parieto-occipital lesion was clearly visualized, adherent to duramater and brain sickle, very hemorrhagic and partly calcified; the small lesion near the Sylvian fissure had the same macroscopic aspect. Corticography was then performed, revealing epileptogenic activity in both occipital and frontal posterior region (near to Sylvian fissure). Using ultrasonic aspiration, we proceeded to disconnection of the entire lesion through a gliotic surrounding plan, without entering the lesion itself. The smallest lesion was first disconnected and then, given the smaller hemorrhagic risk, entirely removed. A fragment of both lesions was biopsied. Histological exam confirmed SWS lesions.
A disconnection surgery was performed in order to minimize the hemorrhagic risk (given the type of lesions) and was successful in the main purpose of extinguish epileptic areas; the whole procedure was made under neuro-stimulation in order to avoid injury to eloquent (motor) areas.

Postoperative course

A distal lower limb left paresia was observed after surgery, with a progressive improvement with physical rehabilitation; there wasn´t a worsening of the previous visual field deficit. With eighteen months of follow-up, the patient is seizure free and despite the minor distal lower limb motor deficit she is completely autonomous.

Discussion

Different surgical procedures are described in the surgical treatment of SWS patients, classic examples being lobectomy or hemispherectomy, as stated above. Desired lesionectomy of SWS patients that have typically large, venous angiomatosis lesions are usually avoided due to the high risk of intra-operative bleeding and subsequent increasing morbidity and mortality.[6] In our patient, a disconnective technique has been developed in order to reduce the above mentioned risks yet maintaining the main goal: seizure control. Disconnective procedures can be defined as an interruption of the epileptic network including discharge-spreading pathway, by isolating the primary epileptogenic area, rather than removing the epileptogenic foci.[6-8] The resection of a small portion of cortical structure is associated with a decrease in short and long term complications. This kind of procedures is in general shorter with a little amount of blood loss.[8-10] However, to achieve success by means of this kind of procedure it is crucial to totally understand the anatomy of the brain and its interlobar/interhemispheric connections.

This technique has helped in this sort of cases, where the epileptic lesions are not well delimited from the surrounding structures or when there are multiple foci responsible for intractable epilepsies[8]. To treat them, some authors concluded that disconnection procedures have a similar success rate to resection procedures with respect to seizure outcome, and with lower morbidity.

In our patient, we decided upon a disconnection technique of the lesion area instead of removing the epileptogenic focus. Our patient had a good outcome: she is seizure free (Engel Class I)[11], with a minimum motor impairment. To our knowledge, there is not any report in the literature considering the disconnective procedure specifically in SWS patients. We believe this surgical technique must be considered for the treatment of refractory epilepsy in selected patients suffering from SWS.

Acknowledgments

To my family and friends and also my mentors who support me.

Disclosure

The authors report no conflict of interest concerning the material or methods used in this study or the findings specified in this paper.

References

1.Comi AM: Presentation, Diagnosis, Pathophysiology, and Treatment of the Neurological Features of Sturge-Weber Syndrome. **The Neurologist**, Volume 17, Number 4, 2011.

2. Brodie MJ, Barry SJE, Bamagous GA, Norrie JD, Kwan P: Patterns of treatment response in newly diagnosed epilepsy. **Neurology, 78**:1548-1554, 1542-1543, 2012.

3.Bodensteiner JB, Roach ES. Sturge-Weber syndrome: Introduction and overview. In: Sturge-Weber Syndrome, 2nd edition, Bodensteiner JB, Roach ES (Eds), Sturge-Weber Foundation, Mt. Freedom, **NJ**, 2010.

4.Maria BL, Neufeld JA, Rosainz LC, et al. Central nervous system structure and function in Sturge-Weber syndrome: evidence of neurologic and radiologic progression. **J Child Neurol** 13:606, 1998.

5.Lee JS, Asano E, Muzik O, et al. Sturge-Weber syndrome: correlation between clinical course and FDG PET findings. **Neurology,** 57:189, 2001.

6. Ribaupierre S, Delalande O: Hemispherotomy and Other Disconnective Techniques. **Neurosurg Focus 25**(3):E14, 2008.

7. Binder DK, Schramm J: Transsylvian functional hemispherectomy: **Childs Nerv Syst 22**:960-966, 2006.

8. Carson BS, Javedan SP, Freeman JM, Vining EP, Zuckerberg AL, Lauer JA, et al: Hemispherectomy: a hemidecortication approach and review of 52 cases. **J Neurosurg 84**:903-911, 1996.
9. Cats EA, Kho KH, Van Nieuwenhuizen O, Van Veelen CW, Gosselaar PH, and PC Van Rijen PC: Seizure freedom after functional hemispherectomy and a possible role for the insular cortex: the Dutch experience. **J Neurosurg 107** (4 Suppl):275-280, 2007.
10. Kossoff EH, Vining EP, Pillas DJ, Pyzik PL, Avellino AM, Carson BS, et al: Hemispherectomy for intractable unihemispheric epilepsy etiology vs outcome. **Neurology 61**:887-890, 2003.
11. Engel, Jerome: Surgical Treatment of the Epilepsies: **Lippincott Williams & Wilkins**, 1993, ISBN 0-88167-988-7.

Figure legends

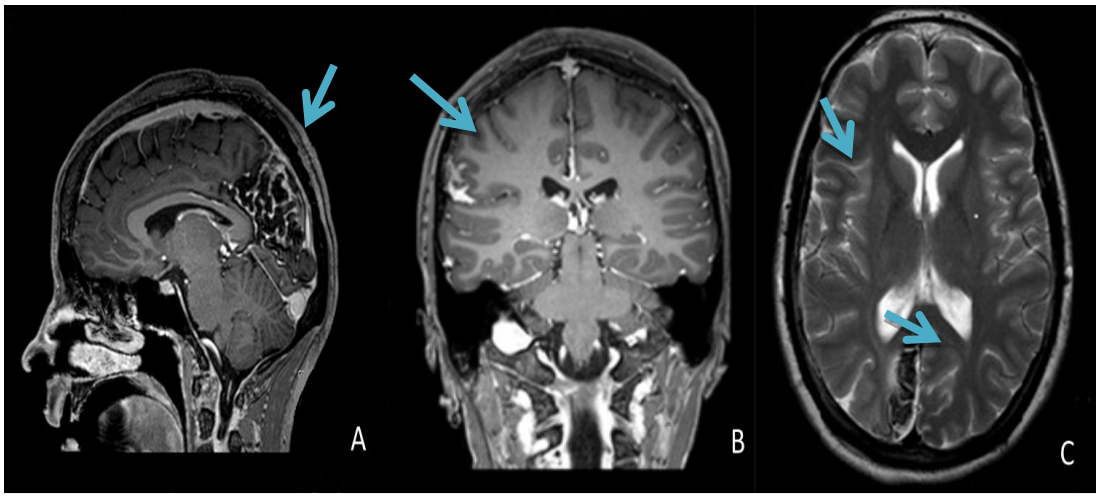


Figure 1- MRI, arrows show the temporo-parieto-occipital calcified lesions before surgery.

MRI- Magnetic resonance imaging

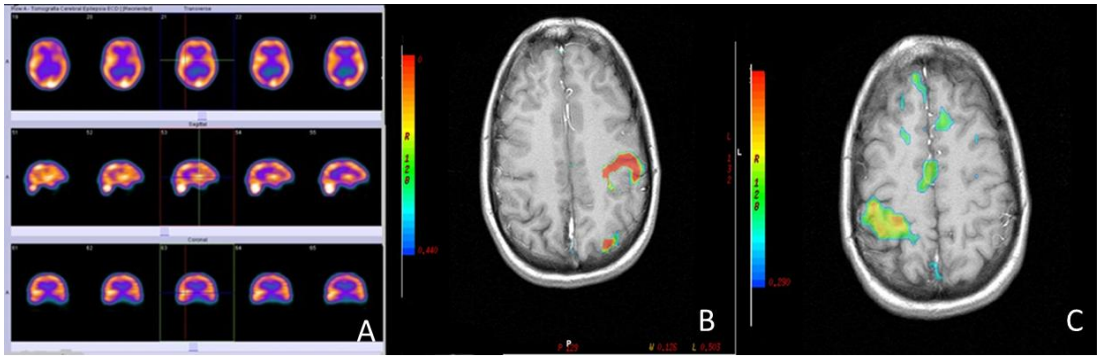


Figure 2- (A) – Ictal SPECT, showing hipoprerfusion mainly in the temporo-parietal-occipital area on the right and left frontal side and also hiperdebits on the right fronto-temporo-parietal areas, suggesting as lesional areas the right parieto-occipital region (lateral to the right parieto-occipital medial one) or right temporal (near to the lateral parietal one, coincident to MRI images); (B) functional MR showing the motor brain activity of the right hand and left hand (C).

SPECT: Single-photon emission computed tomography; MR: magnetic resonance

APPENDIX 1

Resumo do "Case-report"

Desconexão como tratamento cirúrgico eficaz em epilepsias refractárias numa paciente com Sindrome Sturge-Weber

Introdução

Descrita como congénita, a síndrome Sturge-Weber (SSW) é rara e caracteriza-se por malformação dos capilares venosos atingindo não só a pele (manha facial ipsilateral tipo "vinho do Porto"), como também o cérebro (angioma leptomeningeo) e os olhos3 que se associam a anomalias neurológicas e oculares especificas cujo carácter pode ser progressivo. No que concerne às convulsões, uma das principais facetas desta síndrome, acompanham-se geralmente de défices neurológicos focais, normalmente hemiparesia ou defeitos nos campos visuais se o córtex occipital for afectado4. Dada a anatomia anormal dos vasos leptomeningeos, as convulsões ocorrem com uma taxa que ronda os 85%, sendo menos de 50% controladas com fármacos antiepilépticos. Estes são utilizados de modo crónico afigurando-se cruciais no tratamento, no entanto, numa grande percentagem de doentes são ineficazes, designando-se a epilepsia como refractária. Nestes casos, a cirurgia é considerada como a melhor e até mesmo única opção para a eliminação ou controlo das crises convulsivas. Muitas opções cirúrgicas estão descritas para o tratamento desta patologia, sendo os exemplos clássicos a lobectomia ou a hemisferectomia funcional. Defendida por alguns como uma abordagem eficaz, a hemisferectomia, embora recomendada precocemente, comporta algum grau de controvérsia acerca dos pacientes a seleccionar e qual a melhor altura para serem submetidos a este procedimento.1 As complicações inerentes ao procedimento, nomeadamente o elevado risco hemorrágico e os potenciais danos corticais não devem ser menosprezados. Tendo em linha de conta todo este contexto aborda-se de seguida um caso bem-sucedido de uma desconexão cirúrgica numa jovem de 24 anos com epilepsia mal controlada com antiepilépticos causada pela patologia em questão. O resultado foi excelente, o que nos faz acreditar que este procedimento pode ser aplicado ao tratamento de outros pacientes seleccionados.

Caso-clínico:

Jovem de 24 anos, dextra, com o oitavo ano de escolaridade, apresentava-se com uma epilepsia refractária associada ao SSW, seguida na consulta desde 1988.

As convulsões iniciaram quando tinha 2 anos de idade, inicialmente ocorriam num contexto febril com clonias no membro inferior esquerdo com perda parcial da consciência. Nos dias subsequentes, ela manifestava convulsões nos membros esquerdos com duração de 20 minutos sem perda de consciência. Ela referia por vezes uma sensação vertigionosa que precediam as crises e via tudo escuro. Depois disso, apresentava automatismos manuais e movimentos clónicos nos membros esquerdos com perda de consciência. Tinha também hemiparesia pós-ictal esquerda.

Ao exame físico apresentava alopécia na região parietal direita e não apresentava lesões angiomatosas na pele.

Apesar dos diferentes e diversos esquemas terapêuticos instituídos, ela tinha convulsões diárias e muito incapacitantes com várias quedas e diversas lesões durante todos estes anos, pelo que a cirurgia foi uma opção a considerar.

Durante o seguimento da paciente, foram realizados vários exames como EEG inter-críitico e monitorização vídeo-EEG, RM encefálica e funcional, e SPECT ictal e inter-ictal. Dessa vasta avaliação, conclui-se que as crises eram temporo-parieto-occipitais direitas com progressão frontal direita com ocasional generalização secundária. Essa investigação apontou ainda duas possíveis zonas epileptogéneas: parieto-occipital direita e temporal direita.

Os exames campimétricos mostraram uma hemianopsia homónima esquerda.

A paciente submeteu-se a cirurgia em Setembro de 2010. A abordagem utilizada foi a desconexão da área lesional perieto-occipital com remoção da lesão parietal direita adjacente ao vale silviano. De salientar que foi realizada durante a cirurgia a electrocorticografia que evidenciou actividade epileptogénea junto à lesão justa-vale silviana e no lobo occipital direito.

Esta técnica ou abordagem cirúrgica foi realizada de forma a minimizar os riscos hemorrágicos (dado o tipo de lesões) e foi deveras bem-sucedida no seu propósito principal, a extinção das áreas epilépticas. Todo o procedimento foi levado a cabo sob neuro-estimulação de modo a evitar qualquer lesão das áreas eloquentes (motoras).

No follow-up aos 18 meses, foi observada uma parésia distal no membro inferior esquerdo após a cirurgia, com melhoria progressiva com a reabilitação; não houve agravamento no défice prévio dos campos visuais. No decorrer deste período, a paciente

não apresenta qualquer crise e apesar de um défice motor muito discreto no membro inferior esquerdo, ela é completamente autónoma.

Discussão

Diversos procedimentos cirúrgicos estão descritos no tratamento cirúrgico do SSW, sendo os já referidos exemplos clássicos a lobectomia ou a hemisferectomia.

A lesionectomia desejada de pacientes com esta patologia que têm normalmente lesões venosas angiomatosas grandes, são normalmente evitadas devido ao elevado risco de sangramento intra-operatório subsequente aumento de morbi e mortalidade. Neste caso, a técnica de desconexão foi desenvolvida de modo a reduzir os riscos acima mencionados ainda que mantendo o objectivo principal: o controlo das crises. Os procedimentos de desconexão podem ser definidos como uma interrupção da rede epiléptica incluindo a via carga-descarga, pelo isolamento da área epileptogénea primária em vez de remover os focus epileptogéneos.

A ressecção de uma pequena porção de estrutura cortical está associada com a diminuição das complicações a curto e longo prazo. Este tipo de procedimentos é, em geral, mais curto com uma pequena hemorragia. No entanto, para alcançar o sucesso por meio deste tipo de procedimento é fundamental compreender totalmente a anatomia do cérebro e as suas conexões interlobares/inter-hemisféricas.

Esta técnica tem ajudado neste tipo de casos, nos quais as lesões epilépticas não estão bem delimitadas das estruturas envolventes ou quando há múltiplos focos responsáveis pela refractoriedade das epilepsias. Para o seu tratamento, alguns autores advogam que os procedimentos de desconexão têm uma taxa de sucesso semelhante aos procedimentos de ressecção com respeito aos resultados no controlo das crises e à sua menor morbilidade.

Nesta paciente, decidiu-se realizar a técnica de desconexão da área lesional em vez de remover o foco epiléptico. O resultado foi excelente: ausência de convulsões com um prejuízo motor mínimo. Do nosso conhecimento, não há nenhum caso reportado na literatura que considere esta técnica especificamente no tratamento de pacientes com SSW. Acreditamos que este procedimento deva ser considerado no tratamento de epilepsias refractárias em pacientes seleccionados que sofram de SSW.

APPENDIX 2