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Familial Cavernomatosis: The Utility Of Intraoperative Ultrasound - A Case Report

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Abstract
Cavernomas, also known as cavernous angiomas, are small vascular malformations in the brain or spinal cord that resemble a blackberry. They consist of small blood-filled cavities and their growth typically occurs through recurrent microbleeding. Diagnostic imaging, particularly MRI, allows for reliable diagnosis and monitoring, while intraoperative ultrasound could facilitate surgical procedures and consequently improve patient prognosis. We report the case of a 35-year-old patient presenting with drug-resistant generalized tonic- clonic seizures. Surgical excision of the lesion often leads to better seizure control in such patients.
Keywords: *Cavernoma, Familial cavernomatosis, Epilepsy, Cerebral MRI*

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I. Introduction :

Cavernoma is a benign vascular malformation whose pathogenesis is poorly defined. Familial cerebral cavernomatosis is considered rare as it represents about 20% of all cases of cerebral cavernomatosis, with an estimated prevalence between 1/5,000 and 1/10,000. Diagnostic imaging, particularly MRI, allows for reliable diagnosis and monitoring, especially regarding growth and morphological changes in familial cases. The indication for surgical treatment is controversial, particularly in small-sized cavernomas and in functional areas. However, intraoperative ultrasound could facilitate surgical procedures and consequently improve the prognosis of operated patients.

II. Observation :

We report the case of a 35-year-old patient who has been experiencing epilepsy seizures for the past 6 years under treatment. The parents were not consanguineous (the father had previously undergone surgery for a cerebral cavernoma). The patient has been experiencing resistance to anti-epileptic drugs for the past 8 months, without sensory or motor deficits. Neurological examination revealed no meningeal syndrome or signs of focalization.

III. Discussion :

Cerebral cavernomas are characterized by the presence of vascular cavities without intervening neural tissue and surrounded by endothelial walls and thin connective tissue. They lack elastin, muscle fibers, and basement membrane. **(01)**

Their prevalence in the general population ranges from 0.1 to 0.5. The typical age of symptom onset is between 20 and 30 years, although in some cases, symptoms may appear in childhood. Most cases are sporadic and involve a single lesion. The familial form occurs in 10 to 50 cases per series**,(01)**

While familial cavernomas have over 80 cases with multiple lesions. However, there are cases of multiple lesions where both parents are unaffected. **(06)**

La pénétrance clinique et radiologique de la cavernomatose familiale montre de grandes variations inter ou intra familaile **(04) .**

Familial cavernomatosis is an autosomal dominant disease. Three genes have been identified: CCM1/KRIT1 on the long arm of chromosome 7, CCM2/MGC4607 on the short arm of chromosome 7, and CCM3/PDCD10 on the long arm of chromosome 3. **(07)**They account for 53%, 15%, and 10% of familial cavernomatosis cases, respectively. Although these mutations vary greatly, they typically result in a premature stop codon in the messenger RNA coded by the gene**(01).** No mutation has been detected in 22% of cases with multiple cerebral cavernomas.

Surgical treatment of cavernous tumors associated with epilepsy depends on the response and availability of medical treatment. The most evident manifestation in 55% of KRIT1 mutation cases is epilepsy, with 32% detected by cerebral hemorrhage. Although 91% of epilepsy cases are highly medically responsive,

9% remain drug-resistant**(03).** In these cases of refractory epilepsy, surgical excision of the lesion often provides better seizure control than medical treatment alone. Surgical indication remains a delicate question and depends essentially on molecular biology data, lesion location, extent, and association with the presence of venous malformations or pregnancy. **(02)**

The current trend in most neurosurgery teams, especially in children, is towards conservative approaches with uncertain long-term results**(03)**

. Precautions include avoiding sports with a risk of head injury and taking medications with a risk of bleeding (anticoagulants and antiplatelets). **(02)**

IV. Conclusion :

Les cavernomes sont des lésions en évolution constante, avec des changements de taille et de signal. Le risque de saignement ou de resaignement est difficile à apprécier. L'IRM cérébrale (T2*-weighted gradient-écho GRE) est d'une importance considérable dans le diagnostic des cavernomes. Les indications opératoires doivent être conduites avec beaucoup de prudence. La morbidité est plus élevée dans les localisations au niveau du tronc cérébral.

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