Case report

Rasmussen encephalopathy (RE) : A case report .

Abstract

Summary:

Rasmussen's encephalitis (RE) is a rare and severe chronic inflammatory brain disease resulting in drug-resistant epilepsy and progressive hemispheric destruction with neurological deficit. ER is associated with deterioration of background EEG activity, progressive atrophy of the affected hemisphere on MRI and extensive PET hypometabolism over the affected hemisphere. We report the case of a 6-year-old girl who presented with pharmacoresistant epilepsy.

Observation:

A 6-year-old child with no particular medical history presented 6 months prior to admission with epilepsy resistant to all therapeutic classes (antiepileptic drugs). Clinical examination revealed left hemiparesis with facial involvement and constant left hemi-myoclonus.

An EEG showed a pattern compatible with a continuous epileptic encephalopathy that could correlate with Rasmussen syndrome.

Cerebral MRI revealed: right cerebral hemiatrophy with secondary homolateral hippocampal sclero-atrophy, compatible with Rasmussen encephalitis.

The patient received immunoglobulins and a corticosteroid bolus, but without improvement; she was therefore a candidate for surgical treatment.

Rasmussen's encephalopathy is a rare and serious disease that can be responsible for paresis and cognitive decline. Its pathophysiology is still poorly defined, and it mainly affects children.

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Introduction :

Rasmussen's encephalitis (RE): is a rare inflammatory and autoimmune disease with epilepsy characterized by unilateral hemispheric atrophy, associated with drug-resistant focal epilepsy, progressive hemiplegia and cognitive decline. The disease usually affects children and begins with a prodromal phase of mild hemiparesis or infrequent seizures that can persist for up to several years.

It was first described in 1958 by neurosurgeon Theodore Rasmussen as a chronic hemispheric encephalitis causing unilateral atrophy [1].

We report the case of a 6-year-old child who presented with epilepsy resistant to medical treatment for 6 months, in whom the various examinations carried out led to the diagnosis of Rasmussen encephalopathy, enabling us to clarify the clinical, paraclinical and therapeutic aspects of this disease.

Case report :

This is a little girl with the following history: an uneventful pregnancy with good psychomotor development, no notion of consanguinity, no notion of epilepsy disease in 1st-degree relatives, who has been presenting generalized clonic seizures resistant to medical treatment for 6 months (several therapeutic protocols based on antiepileptics have been administered), clinical examination have found persistent left hemicorporeal myoclonus, spastic gait, left hemiparesis and left VII cranial nerve paralysis. The other devices were without abnormalities, and the biological work-up (blood concentration of sodium, glucose, urea and creatinine, calcium and hepatic markers) came back normal.

The EEG revealed: a continuous epileptic encephalopathy that could correlate with Rasmussen's syndrome, supplemented by a cerebral MRI that confirmed this diagnosis by showing: right cerebral hemiatrophy with secondary homolateral hippocampal sclero-atrophy, compatible with Rasmussen's encephalitis.

In terms of treatment, the child had received several families of antiepileptic drugs. On arrival at our hospital, and after further investigations, she was put on bolus of corticosteroids and immunoglobulins, but without clinical improvements; hence the indication for surgical treatment.

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Discussion:

RE is a chronic inflammatory disease responsible for unilateral cerebral atrophy, mainly affecting children but can occur in adolescents and adults, characterized by refractory seizures to treatment and accompanied by progressive loss of neurological function [1]. Annual incidence is estimated at around 2.4 cases/10 million people [1].

The disease evolves in two stages: an acute stage marked by frequent convulsive seizures starting in a single cerebral hemisphere, followed by a residual stage with persistent severe neurological deficits and recurrent epilepsy [1].

RE is an immune-mediated disease, characterized by the constant involvement of lymphocytes. Most of the infiltrating lymphocytes are cytotoxic T lymphocytes. [2]

CD4 T cells are also present, in clonal expansion, and often manifest a Th1 phenotype with secretion of interferon gamma (IFN-γ) and tumor necrosis factor (TNF). It remains unclear whether the antigens triggering this cell-mediated attack are endogenous or reflect a previously undiscovered pathogen (for example a cryptic viral infection). [3]

Humoral hypotheses have been investigated, but humoral mechanisms have not been validated over time, with evidence accumulating in favor of a cell-mediated hypothesis [2].

The diagnosis of RE is based on clinical (focal seizures and unilateral cortical deficit), electrophysiological (EEG slowing and unilateral hemispheric seizures) and morphological (unilateral hemispheric atrophy +/- hypersignal) features, and sometimes on histology (T-cell infiltration with microglial activation) [4].

In our patient, the diagnosis of Rasmussen's encephalitis was made on electroencephalogram and MRI, in association with the clinical presentation of her seizures.

The management of Rasmussen's encephalopathy is multidisciplinary: antiepileptic drugs cannot control seizures, but surgery can. [5]

As epilepsy is due to permanent inflammation, immunosuppressive and immunomodulatory treatments have been used in the hope of helping control seizures, or even slowing or halting disease progression [6]. Studies have mainly consisted of case reports or small series of uncontrolled patients.

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High-dose corticosteroid, immunoglobulin, plasmapheresis and immunosuppressive therapies have all been reported to be more or less effective in controlling seizures and interrupting status epilepticus in patients with RE [7].

Surgery remains the only curative treatment for epileptic seizures in RE, but it has functional consequences, as the only effective surgery is hemispherotomy or hemispherectomy. Homonymous hemianopsia and hemiplegia are inevitable, although both may be present prior to surgery. Hemispherectomy offers one of the best chances of treating epileptic seizures in patients with RE (>70-80% long-term seizure-free outcome)[8].

The decision and timing of surgery must take into consideration the risks and benefits, seizure risk and presumed neuropsychological compromise.

Our patient had received different classes of antiepileptic drugs, corticosteroid boluses and intravenous immunoglobulins, but without improvement, so she was a candidate for surgery.

Conclusion:

RE is a chronic inflammatory brain disease responsible for drug-resistant epilepsy and neurological deficit, our observation focused on a case of RE that did not respond to medical treatment and required surgical management. This is an entity of epilepsy in children responsible for neurological disability, the diagnosis can be difficult at first, management is multidisciplinary, medical treatment with antiepileptic drugs may be insufficient to stop seizures, however immunotherapy and corticosteroid therapy have shown their effectiveness, surgical treatment (such as hemispherotomy) may be the last resort to improve quality of life of the affected child in the absence of contraindication.

References :

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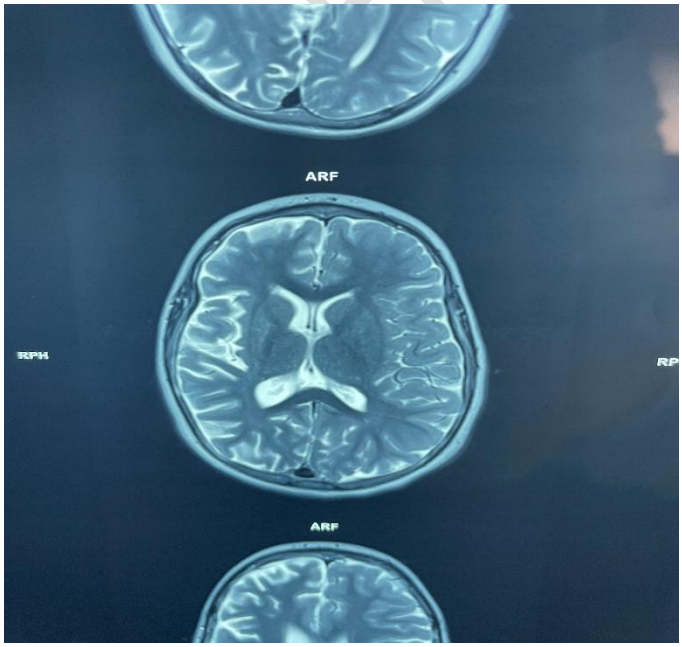

Figure 1

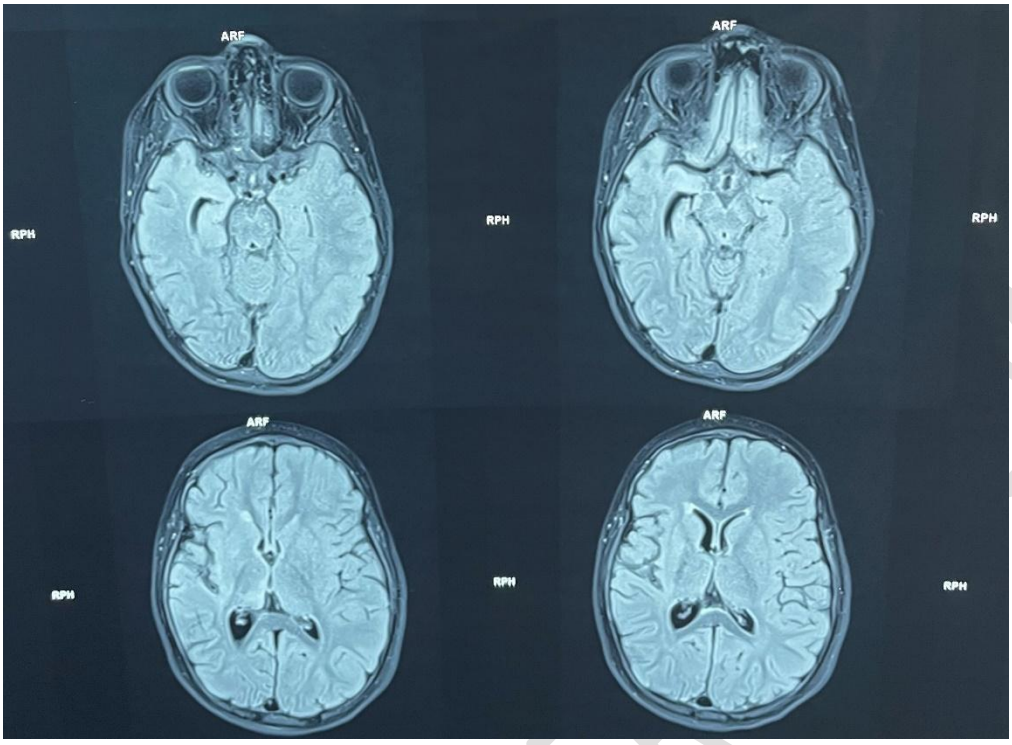

Figure 2

Figure 1(T2) & 2 (FLAIR sequence): showing right cerebral hemiatrophy with secondary homolateral hippocampal sclero-atrophy, compatible with Rasmussen encephalitis.

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