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Progressive Myoclonic Epilepsy how much Difficulty it can cause Before the Final Diagnosis about a case at Fann Hospital in Dakar

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Abstract

Progressive myoclonic epilepsy represents a group of epilepsy characterized by tonic clonic seizures at the start with the association of myoclonus secondarily and intellectual deterioration. This epilepsy poses a problem of diagnosis and management in front of a problem of technical platform.

Keywords: Progressive myoclonic epilepsy; Difficulty

Presentation of the case

16-year-old patient with the notion of second-degree parental consanguinity who, since the age of 14, has presented generalized tonic-clonic seizures preceded by vertigo. First Consultation at the neurology department of CHNU Fann in September 2018.

Neurological examination: normal. The first EEG: generally slowed down, poor trace comprising slow front temporal puffs tending to diffuse she will be put on LP400mg carbamazepine: ½ tab times two per day. With amendment of convulsive seizures.

3 months later: myoclonus occurred; repeated falls associated with atonic seizures and increased seizure frequency

- Neurological examination: dysarthria and cerebellar ataxia
- The second electroencephalography performed poor in sleep patterns with numerous diffuse irritative signs predominantly centro parieto temporal, suggesting symptomatic epilepsy.
- Brain CT and MRI: returned to normal with no particularities.
- Carbamazepine stopped and replaced by gardenal 100mg 1cp / day combined with Depakine 500mg 1cp times two / day. Persistence of seizures despite treatment. In May 2019: State of convulsive illness, which led to his hospitalization in the emergency department. Stabilization of seizures [1,2].

A few days later, she presented secondarily generalized focal motor seizures, she consulted the hospital or a medication instituted whose nature her family ignored with amendment of the seizures an electroencephalography performed: objectified a state of focal electrical sickness interrupted at times by focal crises secondarily generalized motor with a fronto-centro-temporal starting point. On leaving rivotril 2mg: 1cp in the evening was added to the treatment, as well as Keppra and lamictal. In June 2019: she presented a state of epilepticus, which prompted hospitalization at the Albert Royer pediatric hospital in Dakar where she spent a day upon discharge, the neuro examination found: motor aphasia of Broca types, a deficit post-criticism made of a tetraparesis. Of rest and action myoclonus associated with psychomotor regression.EEG: slow sleep of totally desynchronized microarchitecture without physiological figures comprising many diffuse irritative signs compatible with epileptogenic encephalopathy [3-6].

- Stop of Keppra, Gardénal
- Rivotril 2mg: 1 tablet times two per day
- Lamotrigine 100mg: 1 tablet times two per day
- Depakine 500mg: 1 tablet times two per day
- Brain imaging returned to normal.

In view of all these elements, a diagnosis of progressive myoclonic epilepsy was retained.

Introduction

Progressive myoclonic epilepsies are rare, genetically transmitted conditions that are not easy to diagnose early on; and they can be confused in particular with idiopathic epilepsies. They constitute a heterogeneous group of diseases most often family, sometimes sporadic characterized by the occurrence of epileptic seizures, significant myoclonus; the onset marked most often by generalized tonic clonic or clonic seizures, although other seizures such as absence seizures; Mental deterioration progresses to dementia.

Objective: Is it possible to evoke the diagnosis of progressive myoclonic epilepsy without technical means?

Discussion

Clinical elements of diagnosis at the onset phase when there are generalized idiopathic tonic clonic seizures, it can be confused with generalized idiopathic epilepsies. It becomes evident in the course of evolution; and it is then the problem of etiological diagnosis, which arises (Berkovic S), this corresponds to the difficulty noted with our patient. The identification of the causes allow us to take care of our patient in particular by the

genetic test and a sweat gland biopsy. There is a problem of diagnosis due to lack of financial means and the appropriate technical platform. A good assessment of the clinical picture presented by a given patient can immediately point to a specific diagnosis. The field (age of onset, family context, but above all ethnic or geographical origin) is a key element for the diagnosis.

Tonic-clonic or clono-tonic-clonic seizures are constant. Atypical absences, clonic seizures can also be seen in most progressive myoclonic epilepsies. One-sided crises can occur; focal seizures, especially occipital seizures, are characteristic of Lafora's disease (Roger J).Myoclonic syndrome dominates the clinical presentation in most cases, but may remain in the background, behind seizures (early stage) or behind signs of cognitive deterioration or sensory symptoms, especially visual, in the disease of Lafora (Tassinari CA et al), or ceroid-lipofuscinoses. Myoclonus is often maximum in the morning when getting up, interferes (because of its provocation by movement) all the gestures of daily life, including speech, which can become choppy, explosive. They are massive, which can lead to a fall, or segmental and focal, arrhythmic, asynchronous, asymmetric increased by stress, movement or its preparation, especially by the relative rarity of progressive myoclonic epilepsies, which represent less than 1% of the all epilepsies, and their specific clinical presentation, but often confusing (Genton P), means that their diagnosis is sometimes mentioned wrongly, often unrecognized and almost always difficult to confirm [7-9].

Dementia is characteristic of certain etiologies, by its intensity and progression. It is absent in benign adult myoclonic epilepsy (Okino S).

Hierarchy of diagnostic examinations: The diagnosis of the form of progressive myoclonic epilepsy is based on the conjunction of a comprehensive clinical description, a good knowledge of the genetic, ethnic and geographical background and the evolution of symptoms (Genton P). The place of neuropathological explorations with etiological aim has greatly diminished, in particular that of the most aggressive examinations (brain biopsy in particular). In Lafora's disease, the demonstration of starchy bodies is possible in a skin biopsy (Carpenter) carried out in the axillary hollow, rich in sweat glands (the anomalies are particularly visible in the cells of the excretory ducts). Vacuolated lymphocytes can be demonstrated (and analyzed by electron microscopy) in ceroid liposfuscinoses. In ragged –red fibers, the demonstration of “jagged” muscle fibers requires a muscle biopsy, which may however be falsely normal or only slightly altered. A special case is represented by the adult form of ceroid-liposfuscinosis (Kuf's disease), presenting sporadically, with histological damage limited to the central nervous system: a brain biopsy will often be essential for the diagnosis.

Biochemical investigations remain useful in demonstrating enzyme deficits. They turn out to be difficult and often misleading in mitochondrial pathologies as the, the ragged –red fibers (Bindoff LA). Genetic explorations are essential, because they easily confirm the diagnosis, in the disease of Unverricht Lundborg, the dentato-rubropallidoluysal atrophy, juvenile form of Huntington's chorea, ragged –red fibers (Wallace DC), benign familial myoclonic epilepsy in adults (Mikami M), and in certain other conditions [10].

Conclusion

In adolescent epilepsy with generalized convulsive seizures associated with secondary installation of myoclonus and intellectual deterioration. Imaging of which may be normal or cortical atrophy and electroencephalography showing slowing still suggestive of progressive myoclonic epilepsy.

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