Reversible cortical oedema mimicking cortical dysplasia in mitochondrial disorder

Partial seizures are invariably associated with focal brain pathology. Optimised MRI in the evaluation of these patients for surgery has greatly improved the detection of a spectrum of lesions. Imaging findings, however, are not always specific for a particular pathology, and may transiently mimic a fixed structural lesion.

We briefly report the clinical and laboratory findings of a patient who had occipital lobe epilepsy since the age of 18. Seizures consisted of frequent and prolonged auras (hallucinations, palinopsia, photopsia) followed by reversible but protracted visual loss, as well as complex partial and secondary generalised seizures. Clinical findings were short stature, severe hearing and visual loss, mild ataxia, and dysarthria. The patient’s mother had had a stroke at the age of 34 followed by seizures and dementia.

Visual evoked potentials showed abnormal latencies. Monitoring with EEG showed non-specific interictal slowing of background rhythms and focal seizures arising from left and right occipital lobes.

A mitochondrial cytopathy was confirmed by the presence of ragged red fibres and abnormal mitochondrial ultrastructure in the muscle biopsy.

Magnetic resonance imaging during a period of increased seizure activity showed thickening of the cortical ribbon of the right parieto-occipital cortex in T1 weighted images. Increased signal was seen in the T2 weighted sequences (fig 1). A diagnosis of cortical dysplasia was considered and the patient was referred to our centre for surgical evaluation. Repeat MRI five months later no longer showed the lesion (fig 2). Retrospectively, it became apparent that the abnormality was due to transient cortical oedema associated with focal status epilepticus and not a fixed structural pathology of the cortex.

Reversible cortical abnormalities have been shown by MRI in generalised and partial status epilepticus. The appearance may be diagnosed as a neoplasia or ischaemic stroke if the transient neocortical and temporal relation to status epilepticus is not recognised. The unusual linear and pericortical extent of the reversible signal abnormality in our patient led to the initial misdiagnosis of a migrational disorder and the patient was referred for evaluation for surgery for epilepsy. Further investigation showed a mitochondrial disorder in our patient and the transient cortical oedema may indeed be secondary to altered cerebral energy metabolism and the pathogenetic mechanisms causing severe seizures in this condition. This case report illustrates that transient functional MRI abnormalities may mimic fixed structural lesions.

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ABSTRACT

Absence of SCA1 mutation in idiopathic cerebellar ataxia

Idiopathic cerebellar ataxia refers to a group of sporadically occurring cerebellar degenerations of unknown aetiology, which are clinically characterised by progressive ataxia with an onset in adult life. Neuro-pathological and clinical studies suggest that there are at least two types of idiopathic cerebellar ataxia. One group of patients presents with additional non-cerebellar symptoms, such as parkinsonism, autonomic failure, and pyramidal symptoms (idiopathic cerebellar ataxia-P). The underlying pathology in many of these patients is olivopontocerebellar atrophy. Those patients with idiopathic cerebellar ataxia-P who develop severe autonomic failure are subsumed under the broader category of multiple system atrophy. The other group of patients is clinically characterised by a pure...