Facial epilepsia partialis continua in mitochondrial encephalopathy, lactic acidosis, and stroke-like episodes

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A 19-year-old girl presented with continuous left-sided facial twitching movements around the angle of the mouth and cheek for the past 1 month without any abnormal movements of limbs or motor weakness (Video 1). On examination, she was afebrile, her blood pressure was 110/70 mmHg, and pulse rate was 68/min. On neurological examination, she had continuous, left facial movements suggestive of continuous focal seizures, also known as epilepsia partialis continua (EPC). Her past medical history was significant for recurrent headaches, reversible focal deficits mimicking stroke-like episodes, and focal seizures on and off for the past 10 years. For these symptoms, she was thoroughly evaluated and diagnosed with mitochondrial encephalopathy, lactic acidosis, and stroke-like episode (MELAS), corroborated by genetic testing which was positive for MELAS T3243G. There was no history of similar illness in the mother or any other family members. For this recent episode, the patient underwent brain magnetic resonance imaging (MRI), which revealed multiple diffusion-restricted lesions in cortical and subcortical areas with lactate peaks in magnetic resonance spectroscopy (Fig. 1a-f). Her electroencephalography showed intermittent epileptiform discharges consistent with EPC. The patient was started on multiple antiepileptics (levetiracetam 500 mg twice daily, phenytoin 100 mg 3 times a day, and clobazam 10 mg once at night) and anti-oxidants, which provided her with complete relief over a few days.

Mitochondrial disorders can present with a plethora of manifestations [1]. Epilepsy is an associated disorder in certain mitochondrial disorders such as MELAS, myoclonic epilepsy with ragged red fibers, and a few other mitochondrial encephalopathies [2,3]. This has led some researchers to coin the term "mitochondrial epilepsy" to describe these conditions [4]. Cellular hypoxia due to failure of respiratory chain oxidative mechanisms may trigger seizure genesis in the cerebral cortex in these mitochondrial disorders. Seizure generation may also be attributed to reactive oxygen species production, abnormal calcium handling, and increased apoptosis in mitochondrial disorders. But why seizures occur in only a few mitochondrial disorders and not in others is unknown. Rarely, status epilepticus and EPC are also reported in these disorders [5-9]. EPC as such is uncommon, and comprises only 0.1% of all cases of seizure disorder [10]. Mitochondrial disorders are also considered among the important causes of EPC. A plausible mechanism for EPC or status epilepticus is mitochondrial disorders which causes neuronal dysfunction due to persistent energy failure.

Video 1: Left facial continuous jerky movements in a 19 year-old-girl with intermittent epileptiform discharges

The index case earlier had focal seizures, but this time, she had persistent left facial twitching for 1 month, suggestive of the left facial EPC. In EPC, patients have recurrent and, sometimes, unrelenting focal onset seizures with retained awareness occurring over hours, days, or even years as seen in this young girl. MRI brain showed multiple diffusion restriction lesions in the cerebral cortex, showing discrete areas of neuronal dysfunction due to mitochondrial energy failure. This episode may be due to persistent activation of the right motor cortex by one of these new lesions, causing continuous left facial abnormal movements.

This patient responded well to anti-epileptic drugs along with Co-enzyme-Q, L-arginine, carnitine, riboflavin, niacin, folic acid, and thiamine (cocktail regimen), with complete resolution of her facial EPC [11]. She remains symptom-free at 12 months' follow-up. In a young patient with EPC or recurrent focal seizures, mitochondrial disorders need to be suspected, especially MELAS. Brain imaging is helpful in these patients with very specific MRI brain findings of multiple diffusion-restricted lesions in cortical and sub-cortical areas, not following any arterial territories with lactate peaks in magnetic resonance spectroscopy. Furthermore, anti-epileptics and cocktail regimens are helpful in these patients and may lead to complete resolution of the disease.

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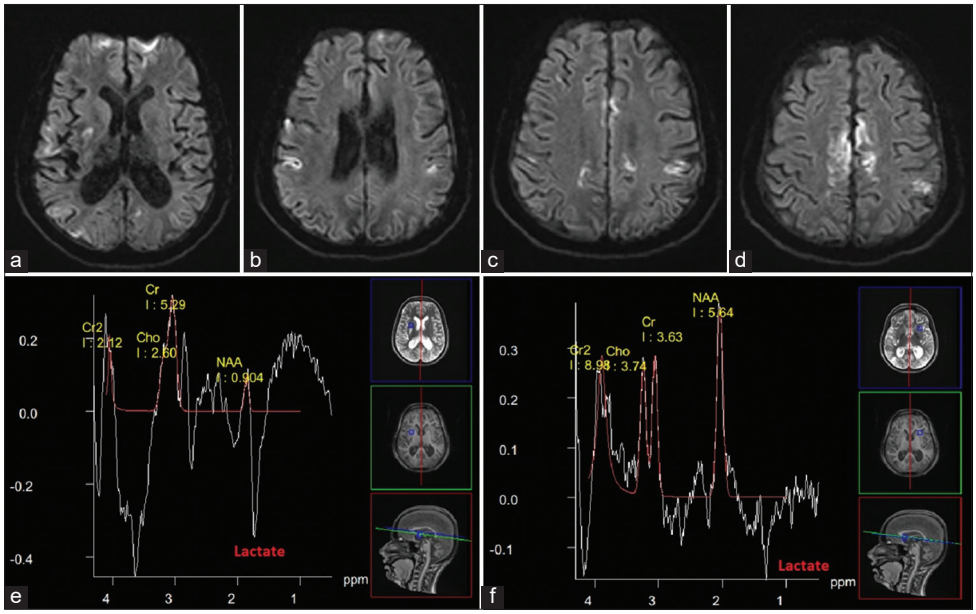

Figure 1: Magnetic resonance imaging, brain showing (a-d) multiple diffusion restricted lesions in cortical and sub cortical areas with (e and f) lactate peaks in magnetic resonance spectroscopy

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