Acute spinal cord lesion in a 5-years-old Leber's Hereditay Optic Neuropathy carrier

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Abstract

Leber's Hereditary Optic Neuropathy (LHON) is classically responsible of optic nerve atrophy; extra-ocular manifestations, known as "Leber plus syndrome", include cerebellar ataxia, Leigh-like encephalopathy and multiple sclerosis (MS)-like syndrome[1]. A large pedigree presented six individuals with maternally inherited LHON due to the G3460A mtDNA mutation; a 5-year-old girl carried the mutation and presented with an acute spinal cord lesion, without any neuropathy.

Case report

The girl presented in Emergency Department after an episode of acute infrascapular back pain occurring without trauma, followed by bilateral legs weakness, and arm weakness within few hours.

On arrival at hospital her vital signs were normal and stable. She was unable to stand and walk with worse weakness in the right leg, her upper extremities were weak, tendinous reflexes were present symmetrically, Babinski sign was present in the right leg. Moreover, she presented impaired bladder control. No touch and pain sensibility alterations seems to be present over the trunk and limbs. No concomitant signs of infection or inflammation were present, neither reported in the previous weeks.

Results

Brain and Spinal cord Magnetic Resonance Imaging (MRI) was performed, showing hyperintense signal alterations in T2-weighted sequences and restricted diffusion in Diffusion Weightened Imaging (DWI) sequences in the anterior portion of spinal cord from C6 to D2, suggesting anterior spinal artery territory involvement. Angio-Computed Tomography (CT) was normal.

Visual and somatosensory evoked potentials were normal.

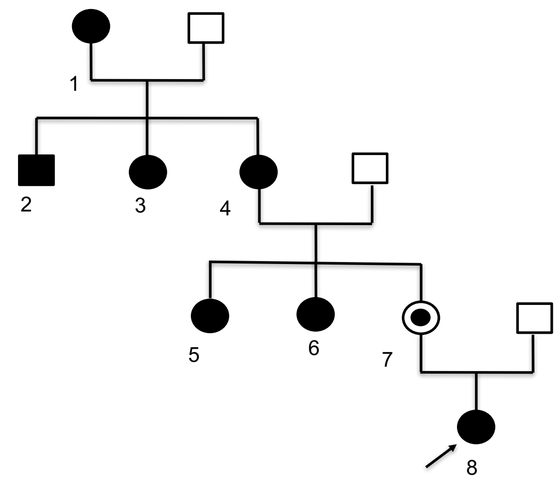
Autoimmunity and thrombofilia screening yielded negative findings. Testing of anti-AQP4 and anti-MOG resulted negative. An **ecocardiography** assessed normal heart and aorta features.

A control spinal cord MRI together with **^{31}Phosphorus-Spectroscopy** was performed 10 days later, showing the complete regression of alterations and no abnormal metabolites.

Aspirin was introduced at low doses (2.5 mg/kg daily), and high dose Methilprednisolone (20 mg/kg daily) for five days were administered.

A treatment with Idebenone was started raging 300 mg three times a day; then continued low dose aspirin. Within three days the clinical manifestations improved. She continued outpatient rehabilitation, and motor ability were normalized within 2 months.

Family history



	1	2	3	4	5	6	7	8
Gender	F	M	F	F	F	F	F	
Mutation G3460A	Heteropl	Homopl	Homopl	Homopl	Homopl	Heteropl	Homopl	
Age at onset	unknown	unknown	unknown	30	21	9	-	5
Signs of onset	Ipovisus	Ipovisus	Ipovisus	Ipovisus Unilateral to bilateral optic neuritis	Ipovisus Unilateral optic neuritis	Ipovisus Bilateral optic neuritis	No signs at ophthalmologic visit 2011	Spinal cord lesion
MRI brain signs	-	-	-	Multiple periventricular subcortical lesion	-	-	-	negative
OCB in CSF	-	-	-	absent	-	-	-	absent
Progression of the disease	Blindness	Blindness Headache	Blindness Psychiatric comorbidities	Vertigo Tremor Diplopia Psychiatric comorbidities	Bilateral scotoma Headache	Monolateral scotoma left eye; blind right eye	-	-
Disorders associated	unknown	unknown	unknown	Rolandic epilepsy Pericarditis ANA pos.	Osteoid osteoma	LLAC pos. connectivitis	Headache	Cutaneous mastocytosis
Therapy						Idebenon	-	Aspirin Corticosteroids Idebenon
Follow-up	20 ys	20 ys	20 ys	20 ys	7 ys	14 ys	5y	1 y

Discussion

Spinal cord lesions are a neurological emergency, with a rare occurrence in the paediatric population and difficult to diagnose[2].

Mitochondrial dysfunctions display a spectrum of manifestation including MS-like and stroke-like lesions, which pathogenesis have not been completely clarified. Stroke-like lesions evolve in an area incongruent to a vascular territory, with a concomitant vasogenic edema[3,4].

Management goals are based on preventing deterioration and secondary events. Corticosteroids, anticoagulation with heparin and antiaggregation with low dose aspirin may provide some benefits, but efficacy has not been documented.

No spinal cord lesions were previously described in literature in LHON. Two cases of spastic paraparesis in Leber's 11778 mt DNA mutated were reported, interpreted as MS-like syndromes.

This case may represent a novel infantile presentation LHON G3460A mtDNA mutation.

References

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