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A Case Report of Leukoencephalopathy with Brain Stem & Spinal Cord Involvement &Elevated (Lbsl) With Enhancement in Cervical Cord Mri

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Case Report:

Presenting case is a 16-year-old woman with a history of febrile convulsion at the age of two with facial palsy and upward gaze. She had delayed motor milestones since the beginning of walking. At 11 years old, she gradually developed weakness of extremities, starting in lower limbs and then the upper limbs and gradually she was unable to walk without assistance. She did not mention sphincter nor sensory abnormalities. The patient goes to exceptional school due to movement problems.

She had no family history of neurological disorders and her siblings were all healthy. She had been treating for Multiple Sclerosis during this period but did not improve.

General examination revealed kyphoscoliosis and generalized muscle atrophy.

On neurological examination, a decrease in bilateral visual acuity with bilateral disc atrophy was observed. She had difficulty speaking. Nystagmus was spontaneous in all directions. Upper extremity tone was spastic and lower extremity tone was flaccid with atrophy and decreased force and bilateral Babinski sing. Sensory testing revealed a distal symmetrical decrease in position and vibration sense.

Blood chemistry, serum ammonia, serum creatine kinase, thyroid function, antinuclear antibodies, serum copper, serum caeruloplasmin, and cerebrospinal fluid testing including CSF protein, IgG level, IgG index, and IgG synthesis were all normal. blood lactate was elevated. A brain MRI (3.0 Tesla) of the brain revealed non-enhancing lesions, hypointense on T1-weighted and hyperintense on T2-weight images. The image characteristics were consistent with the typical LBSL a.in cervical MRI revealed enhancing lesions. MRS study with single and multi voxel method revealed mild increased choline/Cr ratio and increased lactate peak and MI in periventricular white matter no obvious change in NAA is noted. Genetic testing revealed DARS2 mutations in our patient.

Here we report an unusual case of LBSL with enhancement in cervical cord. To our knowledge, there is no reported presentation of enhancements in brain and cervical cord in LBSL

Discussion:

Leukoencephalopathy with brainstem and spinal cord involvement and lactate elevation (LBSL, OMIM # 611105) is an autosomal recessive disease of the central nervous system caused by mutations in *DARS2*[1]. This gene, located on chromosome 1, has 17 exons and encodes the mitochondrial aspartyl-tRNA synthetase[1]. Defects in this gene in neurons impair the translation of mitochondrial mRNAs, leading to mitochondrial dysfunction and progressive cell loss[2]. The disease is characterized by lower limb spasticity, cerebellar ataxia and involvement of the dorsal column[3]. The clinical presentation is variable both in age at onset (early childhood or adulthood) and

in associated features (learning difficulty, epilepsy, mental deterioration and others)[4]. Brain magnetic resonance imaging (MRI) shows diffuse cerebral white matter changes in pyramidal tracts, the brainstem, the cerebellar peduncles, the mesencephalic trigeminal tract, the cerebellar white matter with signal abnormalities in the dorsal column and lateral corticospinal tracts in addition to spectroscopic findings of increased lactate[3,5,6].

Conclusion:

Our report suggests some additional features of LBSL and pathobiological studies are needed to further investigate this disorder.

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