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GALACTOSEMIA AS CAUSE OF WHITE MATTER LESIONS AND TREMOR

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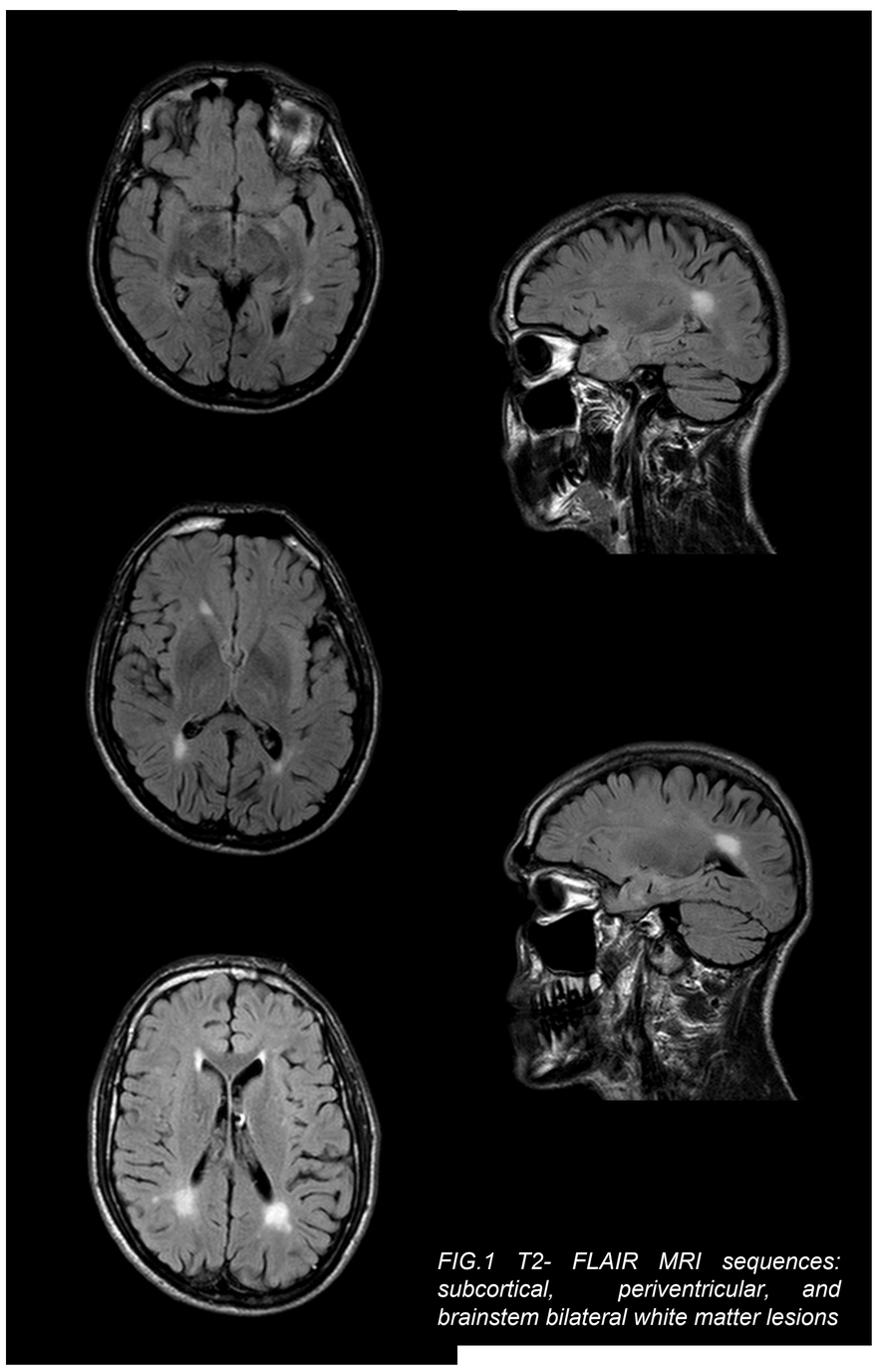

FIG.1 T2- FLAIR MRI sequences: subcortical, periventricular, and brainstem bilateral white matter lesions

Background - Leukoencephalopathy associated with movement disorders represent a diagnostic challenge for clinical neurologists. In subjects with periventricular white matter lesions, suggestive of a demyelinating disease, the relief of a cognitive impairment, together with cerebellar signs and telekinetic tremor in the early stages are unfrequent. A detailed anamnestic research and a wide panel of differential diagnostic analisys should be sistematically adopted.

Case Report - A caucasian 34-year-old man with a history of mild cognitive impairment attribuited to perinatal disseminated intravascular coagulopathy and a referred unspecified malabsorbtion disease with "milk-intolerance" presented at our department because of a 2 years history of tremors. Neurologic examination showed dysarthria, telekinetic tremor, mild dysmetria, diffuse hyperexcitable tendon reflexes, mild hypotonia, ataxo-spastic gait and unstable balance. Neuropsycological evaluation detected psychomotor slowing, mild cognitive impairment, involvement of executive functions and mnesic deficits. He was thin with a BMI of 18. MRI revealed subcortical, periventricular, and brainstem bilateral white matter lesions without Gadolinium enhancement suggestive for a demyelinaiting disease (Fig.1). Evoked potentials were all alterated except for visual one. Cerebrospinal fluid with oligoclonal band research were normal. Standard blood test, tyroid autoantibodies and function, autoimmunity tests, immunoglobulines, borrelia, treponema and epatitis virus antibodies resulted negative.
Genetic for spincerebellar ataxia genes (ATXN1, ATXN2 and ATXN7) rouled out negative. Because of the anamenstic malabsorbtion history, genetic analysis for galactosemia was made and it resulted positive.

Discussion - Classical galactosemia is a rare autosomal recessive inborn error of metabolism (incidence of 1:50000 births) leading to toxic accumulation of galactose and derived metabolites. It usually presents with acute systemic complications in the newborn. Galactose restriction resolves these symptoms, but long-term complications, such as premature ovarian failure and neurological problems occur despite adequate treatment. Neurologic complications including motor dysfunction, speech deficits, cognitive impairment and cerebellar ataxia have been reported in 18-45% of patients and the underlying biochemical mechanism leading to central nervous system damage remain uncertain. Neuroimaging studies confirm poor myelination, scattered white matter abnormalities, cerebral and cerebellar atrophy. Particularity of our case is the presence of a wide spectrum of neurological complications and white matter lesions in a patient probably diagnosed early in life and in not correct diet regimen. Evolutivity of the disorder brought to a complete revalutation of the case with a “re-diagnosis” in adulthood.

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