Genetic Testing and Molecular Biomarkers

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Editorial

A rare autosomal recessive white matter condition known as megalencephalic leukoencephalopathy with subcortical cysts. During the first year of life, both patients experience macrocephaly and demonstrate a gradual loss of motor functions, including ataxia and spasticity. The disorder can be diagnosed using the combined features of magnetic resonance imaging: subcortical cysts and diffusely irregular and slightly swollen cerebral white matter in the anterior temporal region, as well as the front parietal region. Electron microscopic examination of a brain biopsy taken from a patient with MLC showed multiple vacuoles between myelin sheath lamellae, as well as splitting at the intraperiod lines. Mutations in the MLC1 gene are present in approximately 80% of patients with MLC. Despite having a standard MLC phenotype, there is still a subset of patients who have no MLC1 mutations. Owing to increased genetic variability, genetic linkage studies with these families have not resulted in the discovery of another disease locus. Mice lacking the CLCN2 gene, which codes for the chloride channel protein 2 (ClC-2), have been shown to have widespread vacuolation in the brain and spinal cord.

Vacuoles appeared in the central but not the peripheral myelin sheaths. ClC-2 is used in astrocytic end feet lining blood vessels and Bergman glia, much like MLC1. The similarity in white matter defects between MLC patients and homozygous CLCN2 knockout mice, as well as the identical location of the MLC1 and ClC-2 proteins in the brain, made CLCN2 an ideal candidate for a second MLC disease gene. Materials and Procedures MLC1 study and patients This research involved eighteen patients with a standard MLC clinical and magnetic resonance imaging phenotype but no evidence of MLC1 gene involvement. DNA sequencing of genomic DNA and cDNA, quantitative reverse transcriptase-polymerase chain reaction (PCR), and multiplex ligation dependent probe amplification (MLPA) analysis were all used in the MLC1 study (SALSA MLPA KIT P107 for neuro-metabolic disorders [MRC, Holland, Amsterdam, The Netherlands]). Gene Marker was used to interpret the results.

The VU University Medical Center's Institutional Review Board approved the use of patients' materials for further genetic research.

Electrophysiology

Electrophysiology is a branch of biology that deals with the by site directed mutagenesis, the amino acid modification p.Thr396Met was introduced into hClC-2 (cloned in the expression vector pFROG) and confirmed by sequencing. Xenopus oocytes were subjected to a two-electrode voltage-clamp examination, as stated previously.

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