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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: sub cortical 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 peripheric 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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