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Neuro Images

Figure. (A) T2-weighted brain MRI scan. Increased signal involving the white matter. (B) Mitochondrial DNA analysis. Hybridization pattern of Pvu II-digested total DNA from a control (lane 1) and from the patient (lane 2). DNA was extracted from muscle. Asterisks indicate deleted molecules.

MNGIE: Diarrhea and leukoencephalopathy

P. Labauge, MD, PhD, Nîmes, France; R. Durant, MD, Montpellier, France; G. Castelnovo, MD, Nîmes, France; A. Dubois, MD, Montpellier, France

A 47-year-old man had muscular atrophy, cachexia, and chronic diarrhea since age 12. At 33, he presented progressive hearing loss, ophthalmoplegia, and bilateral ptosis. Extended leukoencephalopathy was seen on MRI (figure, A). Muscle biopsy found ragged-red fibers on Gomori-trichrome, staining with cytochrome *c* oxydase negative fibers, and defect in complexes I and IV on mitochondrial respiratory chain analysis. Southern blot analysis revealed multiple mtDNA deletions and a depletion in muscle (see figure, B). Leukocytes thymidine phosphorylase activity

was undetectable. Compound heterozygous mutations (missense mutation in exon 3 (Glu87asp) and single nucleotide deletion in exon 6 (del C2799)) were found in the gene encoding thymidine phosphorylase. Diagnosis of myoneuronal–gastrointestinal encephalopathy (MNGIE) was retained.^{1,2}

Acknowledgment

The authors thank Pr. D. Figarella-Branger (CHU de Marseille), Dr. V. Paquis (CHU de Nice), and Dr. A. Slama (CHU de Bicetre) for their contribution.

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Neurology 2002;58;1862

DOI 10.1212/WNL.58.12.1862

This information is current as of June 25, 2002

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