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Neuro *Images*

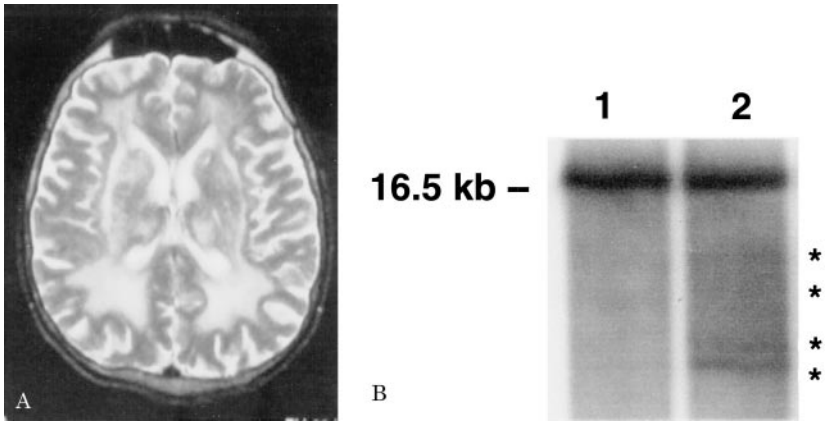

Figure. (A) T2-weighted brain MRI scan. Increased signal involving the white matter. (B) Mitochondrial DNA analysis. Hybridization pattern of Pvu *II-digested total DNA from a control (lane 1) and from the patient (lane 2). DNA was extracted from muscle. Asterisks indicate deleted molecules.*

MNGIE: Diarrhea and leukoencephalopathy

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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]

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