Massive and exclusive pontocerebellar damage in mitochondrial disease and \textit{NUBPL} mutations

A 23-year-old man had progressive nystagmus, cerebellar ataxia, pyramidal signs, and slurred speech since toddlerhood. MRI showed T2 hyperintensity of the cerebellum, the anterior brainstem, and the pyramidal tract, sparing the pontine tegmentum (figure, A). Lack of cerebellar NAA and choline on proton magnetic resonance spectroscopy, glucose hypometabolism on \textsuperscript{18}FDG-PET, and elevated cerebellar lactate suggested mitochondrial disease.\textsuperscript{1,2} Increased cerebellar blood flow suggested vessel proliferation, consistent with mitochondrial disease (figure, C). Skeletal muscle mitochondrial complex I activity was 31\% of control. There was compound heterozygosity for 2 \textit{NUBPL} mutations: c.205\_206delGT, producing a premature stop codon (p.Val69Tyrfs*80); and c.815\_217T/H11022, creating a splicing site and frameshift (p.Asp273Glnfs*32).

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