A 7-year-old boy was referred at age 24 months with failure to thrive, global psychomotor delay, and spastic-ataxic gait with bilateral Babinski sign. Last examination revealed a further psychomotor regression and low IQ. The child could not talk, sit, or walk autonomously and showed upper limb dystonia. His Spastic Paraplegia Rating Scale1 score was 35/52.

Brain neuroimaging showed cerebellar atrophy and bilateral symmetrical hyperintensity in the striatum (figure). Blood lactate levels were slightly elevated. A muscle biopsy showed multiple defects of oxidative metabolism and signs of mitochondrial proliferation. Exome sequencing revealed the homozygous c.553C > T/p.Arg185Trp in PMPCA.
PMPCA processes multiple mitochondrial proteins, including frataxin and iron–sulfur clusters involved in brain energy and oxidative metabolism. Leigh-like neuroimaging and spastic ataxia expand the spectrum of neurologic presentations linked to PMPCA.

Author contributions
Dr. Rubegni: designed and conceptualized study, analyzed the data, drafted the manuscript for intellectual content. Dr. Pasquariello: major role in the acquisition of data, drafted the manuscript for intellectual content. Dr. Dosi: major role in the acquisition of data. Dr. Astrea: major role in the acquisition of data. Dr. Canapicchi: interpreted the data, drafted the manuscript for intellectual content. Dr. Santorelli: interpreted the data, revised the manuscript for intellectual content, provided funds for the study. Dr. Nesti: interpreted the data, revised the manuscript for intellectual content.

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References
Teaching NeuroImages: Leigh-like features expand the picture of PMPCA-related disorders
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