Myoclonic epilepsy with ragged red fibres (MERRF) and mitochondrial encephalopathy, lactic acidosis and stroke-like episodes (MELAS) are established phenotypes of mitochondrial encephalopathies. Nearly all patients affected by MERRF harbour a mutation in the mitochondrial tRNA(Lys) gene. We report a 13-year-old patient who presented with the classical phenotype of MERRF but was found with the typical mutation of MELAS. The patient presented with myoclonic epilepsy beginning at 10 years of age, a muscle biopsy with ragged red fibres and some COX negative fibres and progressive bilateral MRI hyperintensitivities in the basal ganglia constituting MERRF syndrome but lacked clinical characteristics of MELAS. In particular, stroke-like episodes or lactic acidosis were not present. None of the tRNA mutations described in MERRF were found. However, further analyses showed the tRNA(Leu) mutation m.3243A>G usually found in MELAS to be responsible for the condition in this patient. This report highlights the broad phenotypic variability of mitochondrial encephalopathies with juvenile onset. It shows that m.3243A>G mutations can cause classical MERRF and emphasises the significance of comprehensive genetic studies if mitochondrial disease is suspected clinically.