

Simultaneous ^{18}F -FDG PET/MRI Assists Diagnosis of a Rare Disease, MELAS

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Abstract: MELAS (mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes) is a rare congenital mitochondrial DNA mutation disease. Here, we report a 4-year-old girl, who presented with short stature, mental retardation, and recurrent seizures, underwent simultaneous ^{18}F -FDG PET/MRI examination. An interesting contradiction images were found on bilateral frontal, left temporal, occipital, and parietal lobes, which were with high blood flow shown on 3D-ASL perfusion images, but low uptake of ^{18}F -FDG on PET images. The contradiction of high blood flow and low glucose metabolism gave us a clue to make the diagnosis of MELAS. The final diagnosis was MELAS confirmed by genetic testing.

Key Words: PET/MR, ^{18}F -FDG, MELAS, diagnosis, simultaneous

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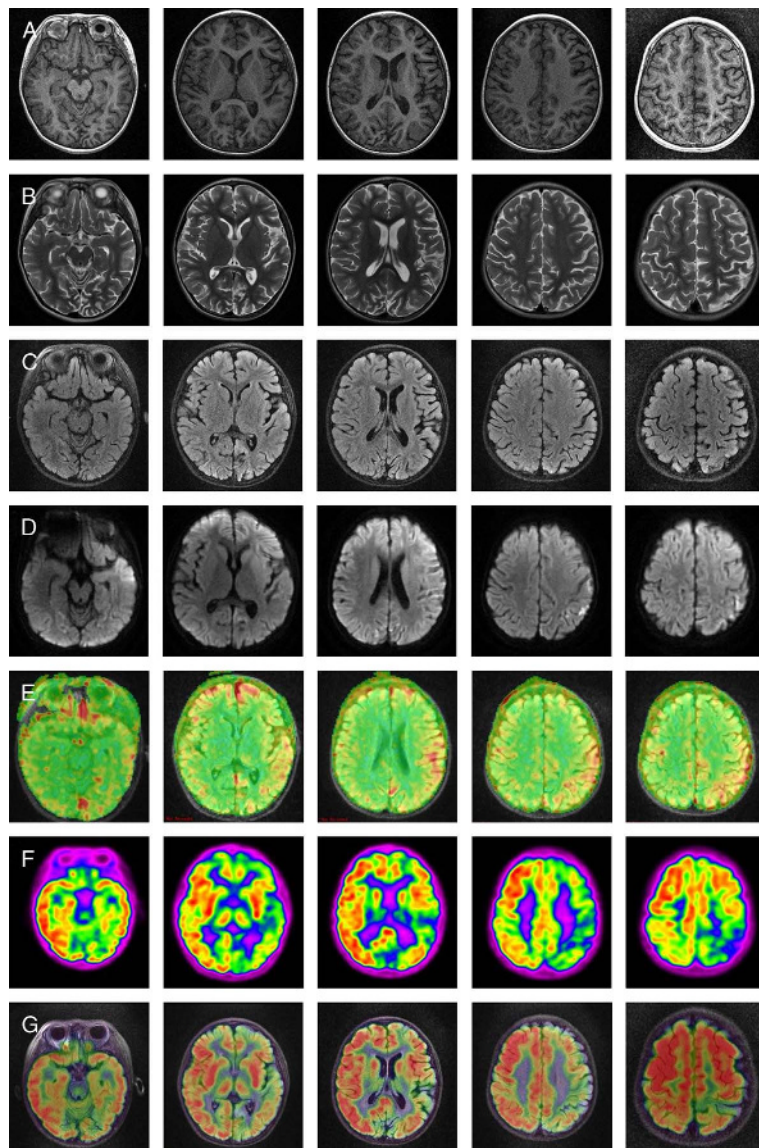


FIGURE 1. MELAS (mitochondrial encephalomyopathy, lactic acidosis, and stroke-like episodes) is a rare congenital mitochondrial DNA mutation disease. A 4-year-old girl, who presented with short stature, mental retardation, and recurrent seizures, underwent ^{18}F -FDG PET/MRI examination. **A–G**, Images from left to right, represent the same level images from base to top of the skull. Diffused atrophy with widening sulci was shown on T1- (**A**) and T2-weighted images (**B**). Hyperintensity was found in bilateral frontal, left temporal, occipital, and parietal lobes on T2 flair images (**C**). DWI showed diffusion restricted in the left temporoparietal lobe (**D**). 3D-ASL perfusion images showed increased cerebral blood flow in the regions consistent with the hyperintensity on T2-flair images (**E**). However, low ^{18}F -FDG uptake was noticed in bilateral frontal lobe, left temporal, occipital, and parietal cortex on PET images (**F**) and PET-T2 flair fused images (**G**), which was contradicted with the hyperperfusion shown on 3D-ASL perfusion images (**E**). The contradiction of high blood flow and low glucose metabolism gave us a clue to make the diagnosis of MELAS. The final diagnosis was MELAS, which was confirmed by genetic testing with the result of **A** to **G** translation at nucleotide 3243 of mitochondrial DNA. MELAS is a rare maternal inherited diseases mainly caused by m.3243A>G mutation within the mitochondrial DNA (mtDNA)-encoded tRNA leucine 1 (*MTTL1*) gene.¹ This patient was detected m.3243A>G mutation 100% loaded in urine and cerebral spinal fluid, and 72.7% loaded in blood. MELAS has multisystem symptoms with onset typically in childhood. The common triad symptoms are lactic acidosis, seizures, and stroke-like episodes, other symptoms include short stature, sensorineural hearing loss, diabetes, cardiomyopathy, and dementia.^{2,3} The point mutation in mtDNA induces the metabolic disorders at cellular level, which leads to generate reactive oxygen species, increase oxidative stress, and damage surrounding cells.^{4,5} Furthermore, it leads to endothelial dysfunction of cerebral blood vessels, which corresponds with cerebral hyperperfusion, vasodilatation, and vasogenic edema in acute lesions. In contrast, the glucose metabolism decreases due to the impaired mitochondrial and respiratory dysfunction.^{6,7} Simultaneous PET/MRI is an important imaging modality to display the abnormalities of MELAS. ^{18}F -FDG PET shows the glucose metabolism decreased on the lesion, and MRI provides diagnostic information of fine-scale structural and functional properties and changes in the brain.⁸ Thus, hybrid PET/MRI contributes to a better understanding of the disease pathology.