

Brain single photon emission computed tomography in patients with A3243G mutation in mitochondrial DNA tRNA.,

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摘要

Abstract

Brain single photon emission computed tomography (SPECT) studies were conducted in three patients with A3243G mutation of the mitochondrial (mt) DNA tRNA. All were born to mothers suffering from chronic progressive external ophthalmoplegia (CPEO) with the same A3243G point mutation of the mtDNA tRNA. The first case manifested clinically with MELAS, the second case manifested with CPEO, and third case was characterized by recurrent migraine-like headache, tremor, and epilepsy. Brain SPECT of all patients, regardless of whether they had or had not suffered from stroke-like episodes, showed multiple areas of asymmetrical decreased perfusion, particularly in the posterior and lateral head regions, especially the temporal lobes. Crossed-cerebellar diaschisis may occur. Conventional brain magnetic resonance images failed to show some of the lesions. Decreased regional cerebral blood flow, rather than previously proposed hyperemia, is likely to be the cause. We conclude that mitochondrial vasculopathy with regional cerebral hypoperfusion may be seen on brain SPECT in patients with mitochondrial disorders and A3243G mutations, regardless of whether they have or have not suffered from stroke-like episodes.