Specific structuro-metabolic pattern of thalamic subnuclei in fatal familial insomnia: A PET/MRI imaging study

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Abstract

Background: Dysfunction of the thalamus has been proposed as a core mechanism of fatal familial insomnia. However, detailed metabolic and structural alterations in thalamic subnuclei are not well documented. We aimed to address the multimodal structuro-metabolic pattern at the level of the thalamic nuclei in fatal familial insomnia patients, and investigated the clinical presentation of primary thalamic alterations.

Method: Five fatal familial insomnia patients and 10 healthy controls were enrolled in this study. All participants underwent neuropsychological assessments, polysomnography, electroencephalogram, and cerebrospinal fluid tests. MRI and fluorodeoxyglucose PET were acquired on a hybrid PET/MRI system. Structural and metabolic changes were compared using voxel-based morphometry analyses and standardized uptake value ratio analyses, focusing on thalamic subnuclei region of interest analyses. Correlation analysis was conducted between gray matter volume and metabolic decrease ratios, and clinical features.

Result: The whole-brain analysis showed that gray matter volume decline was confined to the bilateral thalamus and right middle temporal pole in fatal familial insomnia patients, whereas hypometabolism was observed in the bilateral thalamus, basal ganglia, and widespread cortices, mainly in the forebrain. In the regions of interest analysis, gray matter volume and metabolism decreases were prominent in bilateral medial dorsal nuclei, anterior nuclei, and the pulvinar, which is consistent with neuropathological and clinical findings. A positive correlation was found between gray matter volume and metabolic decrease ratios.

Conclusion: Our study revealed specific structuro-metabolic pattern of fatal familial insomnia that demonstrated the essential roles of medial dorsal nuclei, anterior nuclei, and pulvinar, which may be a potential biomarker in diagnosis. Also, primary thalamic subnuclei alterations may be correlated with insomnia, neuropsychiatric, and autonomic symptoms sparing primary cortical involvement.
