論 文 要 旨

Correlation between clinical and radiologic features of patients with Gerstmann-Sträussler-Scheinker syndrome (Pro102Leu)

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Background and purpose: Gerstmann-Sträussler-Scheinker syndrome is a rare hereditary neurodegenerative disorder with clinical heterogeneity. This study is aim to demonstrate the clinical spectrum and radiologic characteristics of patients caused by Pro102Leu mutation in PRNP.

Materials and methods: We retrospect clinical manifestations of five patients from four Japanese families, and comprehensively analyzed their brain MRI, SPECT (N-isopropyl-p-[123I] iodoamphetamine), and PET (18F-2-fluoro-2-deoxy-D-glucose) images.

Results: All patients developed ataxia of lower limbs and trunk, gait disturbance, dysesthesia in legs, and lower limb hyporeflexia. In the early clinical stage before dementia began, no noticeable abnormalities could be observed from brain MRI, but SPECT and PET revealed mosaic-like pattern of blood flow and glucose metabolism of the brain. Predominant abnormalities were found in the occipital and frontal lobes on SPECT and PET analysis, respectively. In SPECT analysis, blood flow of the anterior cerebellar lobes was lower than that of the posterior cerebellar lobes.

Conclusions: Clinical symptoms resulting from failure of dorsal horn of spinal cord and spinocerebellar tracts were observed in all cases. Radiologic findings revealed individual differences of involved region in their brain, which could produce clinical diversity. We identified a downtrend of blood flow in the anterior cerebellar lobes, a projection field of the spinocerebellar tracts, which is an important feature of Gerstmann-Sträussler-Scheinker syndrome.