Post-genomic study on Endothelial Cell of Moyamoya Disease

Kiyohiro Houkin, M.D., Ph.D.

Department of Neurosurgery
Hokkaido University Graduate School of Medicine

When we take a look at its history, research on moyamoya disease has progressed remarkably in the past several decades, from the viewpoints of infection, auto-immune disorder, environmental disorder and genetic abnormality. In these days, genomic study based on familial occurrence has opened the door to RNE213 and other promising gene abnormality using Microsatellite analysis and SNF analysis. Indeed, many new facts concerning the epidemiology of the disease have been revealed and surgical treatments have been drastically improved. However, despite extensive research, the mechanism of moyamoya disease is still unknown. For further clarification of its etiology, innovative studies are therefore indispensable.

Recently, our laboratory found that the $CD34^+$ $VEGFR2^+$ cells were closely involved in the intimal thickening of the supraclinoid internal carotid artery collected from adult patients with moyamoya disease.[60] This study was interesting in a point that a certain progenitor cells also participated in the progressive occlusive lesion in moyamoya disease. However, the roles and the identity of such cells were still unknown.

Currently, several genetic abnormalities are considered to offer the most probable hypothesis. In addition, interesting papers have been presented on the role of the endothelial progenitor cell on the pathogenesis of moyamoya disease. Intuitively, however, it appears that a single theory cannot always explain the pathogenesis of this disease adequately. In other words, the complex mechanism of several factors may comprehensively explain the formation of moyamoya disease. “Double hit hypothesis” is probably the best explanation the complicated pathology and epidemiology of this disease.

The aim of this study is to culture i-PS cell line of moyamoya patients and induce the endothelial cells and establish the pathogenesis of moyamoya disease as a milestone in the direction of its true solution.