Genetic analysis of RNF213 variant in Moyamoya disease

Moyamoya disease is an idiopathic cerebrovascular disorder that is characterized by progressive occlusion of the internal carotid artery (ICA) and its main branches within the circle of Willis. This occlusion results in the formation of a collateral vascular network (the moyamoya vessels) at the base of the brain. “Moyamoya” means “puff of smoke” in Japanese as the appearance of collateral vascular network, the “moyamoya” vessels on an angiogram looks like a puff of cigarette smoke drifting in the air.

Moyamoya disease is known for its high prevalence in East Asian population, and it is known also for its high incidence of familial onset. Recently, a susceptibility gene for moyamoya disease (MMD) was identified in the East Asian population. Several studies revealed high frequencies of the same single nucleotide variant, c.14576G>A (p.R4859K) variant of a gene called ring finger protein 213 (RNF213) in moyamoya disease patients. Our study group has reported that this RNF213 p.R4859K variant is also associated with various phenotypes of intracranial major artery stenosis not diagnosed as moyamoya disease (Stroke 2012, 2013).

In the presentation, genetic feature of moyamoya disease and the clinical importance of RNF213 will be presented. Molecular biological function of RNF213 will also be discussed.