講演情報

一般演題口演セッション

[O-02] 一般演題口演セッション02　【英語】

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座長:北川 一夫(東京女子医科大学脳神経内科), 金澤 雅人(新潟大学脳研究所臨床科学部門脳神経内科学分野)

[O-02-1] RNF213 polymorphism predicts long-term progression and prognosis of intracranial artery stenosis

◦岡崎 周平[1,2], 吉本 武史[2], 渡辺 光太郎[1], 大原 真理子[1], 権 泰史[1], 藤堂 謙一[1], 佐々木 勉[1], 猪原 匡史[2], 望月 秀樹[1]
(1.大阪大学大学院医学系研究科 神経内科学, 2.国立循環器病研究センター 脳神経内科)

[Objective] Intracranial artery stenosis is the predominant etiology of ischemic stroke in East Asia. We examined the association of the *RNF213* p.R4810K variant with the progression and prognosis of intracranial artery stenosis in a 15-year follow-up study. **[Methods]** We recruited patients with intracranial artery stenosis who underwent follow-up MRIs at intervals of >5 years between 2006 and 2021 at two large stroke centers in Japan. Patients diagnosed with moyamoya disease at the time of initial MRI were excluded. The degree of stenosis was evaluated by visual inspection for predefined arterial segments by two blinded observers (kappa = 0.75). Genotyping was performed using a GTS-7000 system. **[Results]** Among 53 enrolled patients, 22 (42%) had the p.R4810K variant. Mean follow-up duration was 10.1±2.7 years ranging from 5.3 to 14.8 years. There was no significant difference in the baseline characteristics and follow-up duration between the variant carriers and non-carriers. Progression of intracranial artery stenosis was observed in 65% of variant carriers and 29% of non-carriers (OR 5.03; 95%CI 1.54 - 16.44; P = 0.010). In a cox regression model adjusted for age and sex, the *RNF213* p.R4810K variant was an independent predictor of the progression of intracranial artery stenosis (adjusted RR 3.52, 95%CI 1.47 - 8.44, P = 0.005) and the development of stroke or transient ischemic attack (adjusted RR 4.18, 95%CI 1.29 - 13.52, P = 0.017). **[Conclusion]** The *RNF213* p.R4810K variant is a strong predictor of long-term progression and prognosis of intracranial artery stenosis.