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Clinical manifestations and a *RNF213* p.Arg4810Lys variant in pediatric patients with hypertension and Moyamoya disease

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Objectives: Moyamoya disease (MMD) -a steno-occlusive disease of the distal cerebral arteries- is often accompanied by hypertension, and *RNF213* p.Arg4810Lys is known as a susceptibility gene of MMD. There is a report that a polymorphism of *RNF213* is associated with systolic blood pressure. The purpose of this study was to evaluate the clinical characteristics and gene study for *RNF213* of Korean pediatric patients with hypertension and Moyamoya disease.

Methods: A retrospective analysis of medical records in 44 hypertension cases (3.9 %) referred to department of pediatrics due to hypertension among 1,137 MMD patients from January 2000 to June 2018 were performed. The presence of renovascular hypertension was confirmed by computer tomography (CT) angiography.

Results: There were 18 girls and 26 boys, and the mean age at the diagnosis of hypertension was 8.9 years. On CT angiography, it revealed bilateral renal artery stenosis of ostial lesions in 8 of the patients and unilateral lesions in 5 of the patients. In 11 patients, the diffuse narrowing of the aorta- especially in the intra-renal abdominal aorta- was detected, and both the narrowing of aorta and renal artery stenosis was combined in 2 patients. In 22 patients, there was no evidence of renal artery stenosis, and they were considered as presumed essential hypertension. The genetic study for *RNF213* gene was performed in 27 patients. The molecular testing revealed homozygosity of *RNF213* p.Arg4810Lys in 13 patients, in whom 12 patients showed the renal artery stenosis or aorta narrowing. Among the 13 patients with heterozygosity of *RNF213* p.Arg4810Lys, 7 patients were compatible with renal artery stenosis or aorta narrowing. In one patient without mutation, there was no evidence of renovascular hypertension.

Conclusions: Our study suggests that *RNF213* may be the causative gene in vasculopathy associated hypertension-renal artery stenosis or narrowing of aorta-in Korean children with MMD.