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Percutaneous coronary intervention in a patient with homozygous RNF213 variant

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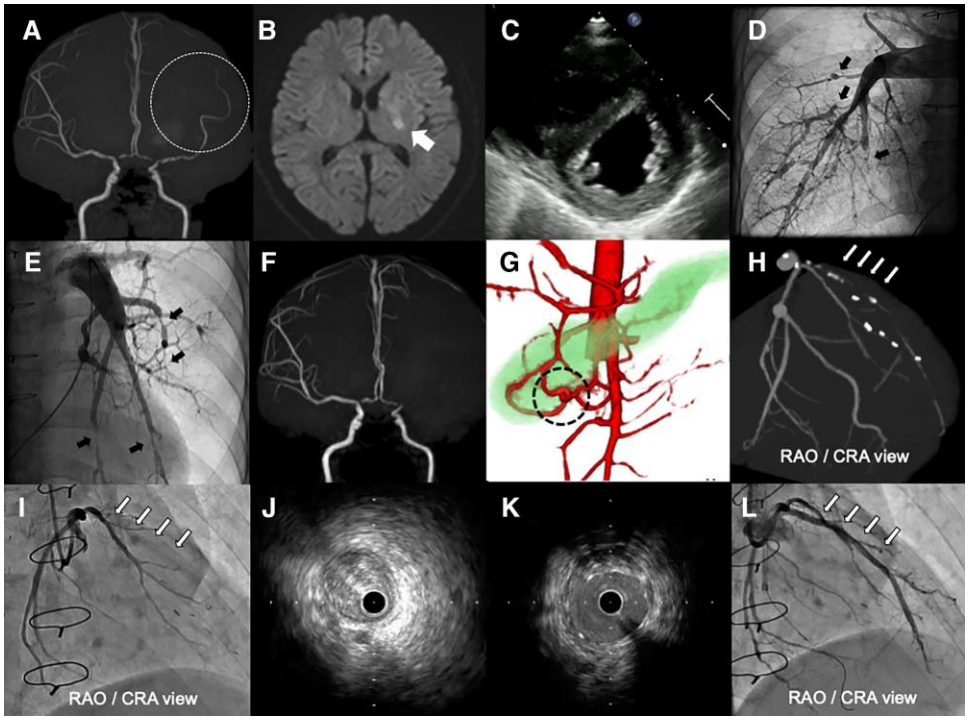

A 44-year-old man with a history of ischaemic stroke was diagnosed with Moyamoya disease with stenosis of the left middle cerebral artery (MCA) (*Panels A and B*). He had complete total occlusion (CTO) of the left anterior descending artery (LAD), peripheral pulmonary artery stenosis (PPS) with pulmonary hypertension (*Panels C–E*), and left internal thoracic artery stenosis. The left MCA stenosis progressed rapidly within a year (*Panel F*) and asymptomatic pancreaticoduodenal artery aneurysms were detected (*Panel G*). Trio whole-exome sequencing identified a homozygous c.14576G > A (p.Arg4859Lys) variant of RNF213. The heterozygous variant was detected in the patient's brother and parents. This variant is found in ~80% of East Asian patients with Moyamoya disease and is associated

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with extracranial involvement, including PPS.[1] Due to the genetic background of systemic vasculopathy, coronary artery bypass grafting for the LAD was performed using the right internal thoracic artery instead of percutaneous coronary intervention (PCI), but occlusion occurred within a month (*Panels H and I*). Therefore, the patient underwent PCI. Intravascular ultrasound imaging revealed that the intramural plaque was eccentric and soft without calcification. Stents were implanted at nominal pressure to reduce vessel injuries by excessive dilation. The plaque was adequately compressed and the stents were fully expanded (*Panels J–L*). Six months later, coronary angiography showed no stent restenosis.

RNF213 vasculopathy involves multiple organs and is progressive. To our knowledge, this is the first report of successful PCI and stent patency for CTO in a patient with a homozygous RNF213 variant. These findings contribute to uncovering the aetiology of and planning treatment strategies for RNF213 vasculopathy.

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Reference

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