Genetic study uncovers mutation associated with fibromuscular dysplasia

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Understanding of fibromuscular dysplasia (FMD), a rare blood vessel disease, is making the jump from the laboratory to the clinic with new findings about a genetic variant.

Researchers found the mutation in a gene that is associated with classical Ehlers-Danlos Syndrome as well, in multifocal FMD. That means it could help clinicians understand whether a person inherited the disease from a relative or another mechanism, in affected families.

"We identified four independent families with the same genetic variant in COL5A1 and vascular disease in a pattern of dysplasia-associated arterial disease, including arterial dissections and multifocal FMD," says senior author Santhi Ganesh, M.D., an associate professor of internal medicine and human genetics, and a cardiologist at the Michigan Medicine Frankel Cardiovascular Center. "Notably, the variant appears to have been inherited from a shared ancestral founder."

Ganesh says the implication of this finding is that other carriers of this variant may exist in the population. The pattern of arterial involvement among carriers of the COL5A1 "G514S" variant is unique, providing clinicians with clues for when to suspect its involvement.

"The identified genetic variant meets clinical criteria for pathogenicity—a first for FMD," she says.

Further, additional variants in the COL5A1 gene were associated with a higher rate of arterial dissections among individuals with multifocal FMD.

More information: Julie Richer et al, Novel Recurrent COL5A1 Genetic Variant Is Associated With a Dysplasia-Associated Arterial Disease Exhibiting Dissections and Fibromuscular Dysplasia, *Arteriosclerosis, Thrombosis, and Vascular Biology* (2020). DOI: 10.1161/ATVBAHA.119.313885