Can Ehlers-Donlas Syndrome Affect the Vascular System

Introduction:
Ehlers-Danlos Syndrome is a clinically and genetically heterogeneous group of disorders that result from some defect in the synthesis or structure of fibrous collagen.(1)

Causes:
Vascular Ehlers-Danlos Syndrome is autosomal dominant, typically caused by a change (mutation) in the COL3A1 gene.(2) The mutation is described includes two types of mutation in COL3A1, a substitution of glycan with amino acids in the triple helix region, which would cause vascular EDS. (4)

Clinical manifestations:
• Skin and joint hypermobility.
• Easy bruising
• Arterial or vascular rapture
• Small joint hyper extensibility (1)
• Gastrointestinal proliferation(4)

Conclusion:
The vascular EDS results from abnormalities in type III collagen, it is autosomal dominant, and results from mutation in structural protein.(3)

Reference:
1. Robbins and Cotran pathologic basis of disease (8th edition)
2. Vascular Ehlers-donlas syndrome / genetic and rare disease information canter (GARD)
4. Genetic aspects of the vascular type pf Ehlers-danlos syndrome – j-stage