



CAN EHLERS-DONLAS SYNDROME AFFECT THE VASCULAR SYSTEM



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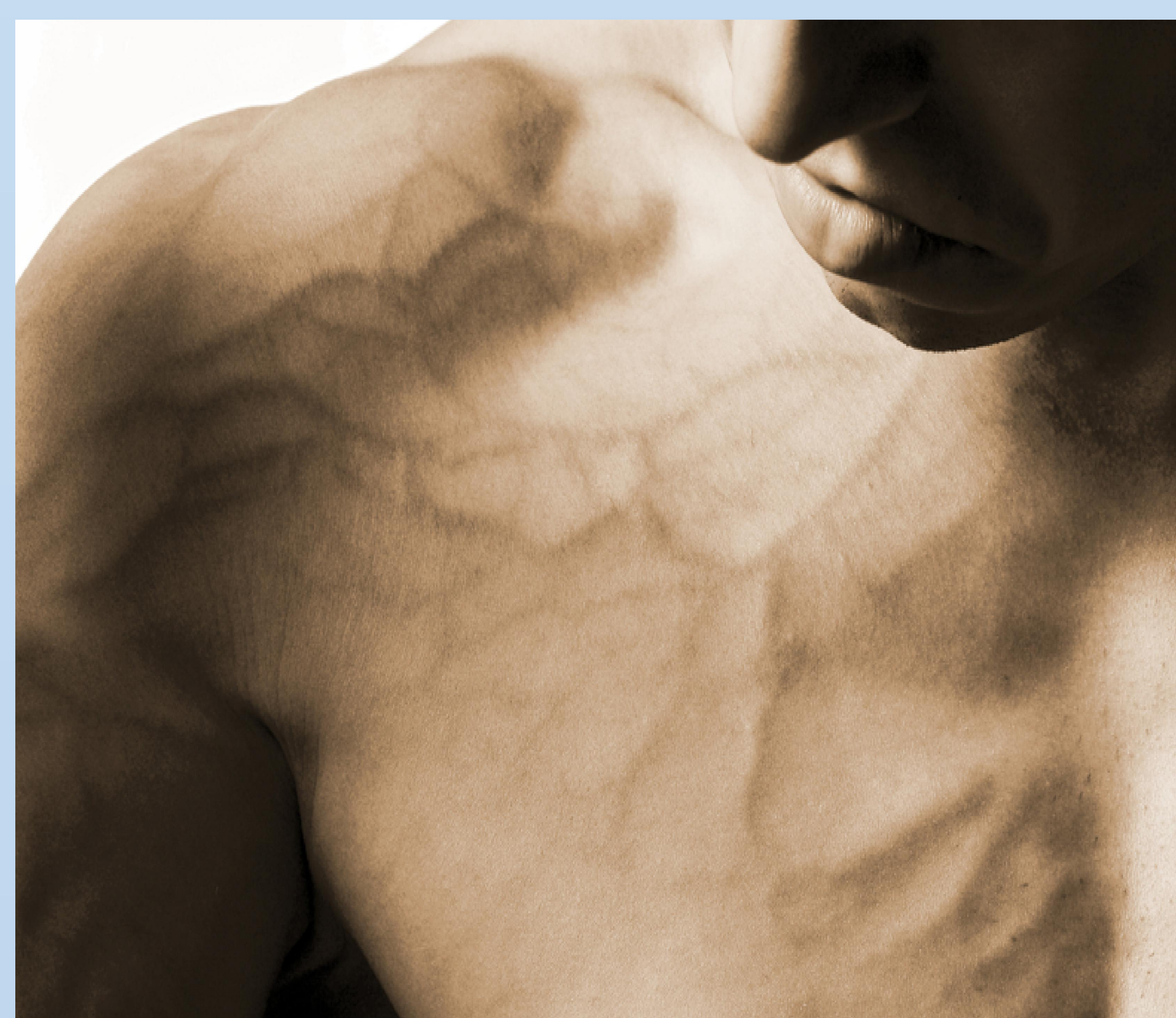
Introduction:

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)



Vascular Ehlers-Donlas syndrome



A hallmark feature of Hypermobile type

Clinical manifestations:

- Skin and joint hypermobility.
- Easy bruising
- Arterial or vascular rupture
- 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. Vasclar Ehlers-donlas syndrome / genetic and rare disease information canter (GARD)
3. Pepin MG, byers PH: Ehlers-Donlas syndrome, vascular type, gene rev.
4. Genetic aspects of the vascular type pf Ehlers-danlos syndrome – j-stage