

DIAGNOSTIC CRITERIA FOR THE VASCULAR TYPE OF EDS (EDS IV)

Patient information	
<i>Date:</i>
<i>NAME:</i>
<i>First name:</i>
<i>Date of birth:</i>
<i>City:</i>
Informed consent:	
<i>I confirm that my clinical data presented here can also be used for medical research and scientific publications without my name (anonymous)</i>	
<i>Signature:</i> <i>(patient or parent/legal representative when applicable)</i>
<i>Place/date:</i>
<i>Motivation:</i>
<i>Referring physician:</i>
<i>Adress of referring physician:</i>
<i>Referring department:</i>
<i>Phone:</i>
<i>Fax:</i>
<i>E-mail:</i>
<i>Material:</i>	<i>blood:</i> <input type="checkbox"/> <i>DNA:</i> <input type="checkbox"/> <i>fibroblasts:</i> <input type="checkbox"/>

The Vascular Type of EDS (EDS Type IV, MIM 130050)

Diagnostic Criteria

The vascular type of EDS is inherited as an autosomal dominant trait, and is caused by structural defects in the pro α 1(III) chain of collagen III encoded by *COL3A1*. It has the worst prognosis, is not so rare as usually considered, and is characterized as follows:

Major diagnostic criteria

- Thin, translucent skin (visible venous pattern over the chest) yes no
- Arterial/intestinal/uterine fragility or rupture yes no
- Extensive bruising yes no
- Characteristic facial appearance (thin nose, small lips, starring eyes, hollow cheeks, missing ear lobe) yes no

Minor diagnostic criteria

- Acrogeria (old looking hands and feet) yes no
- Hypermobility of small joints yes no
- Tendon and muscle rupture yes no
- Talipes equinovarus (clubfoot) yes no
- Early-onset varicose veins yes no
- Arteriovenous, carotid-cavernous sinus fistula yes no
- Pneumothorax/pneumohemothorax yes no
- Gingival recession yes no
- Positive family history, sudden death in (a) close relative(s) yes no

Note: The presence of any two or more of the major criteria is highly indicative of the diagnosis, and laboratory testing is strongly recommended.

Recommended Literature

Beighton P, De Paepe A, Steinmann B, Tsipouras P, Wenstrup RJ (1998): Ehlers-Danlos syndromes: revised nosology, Villefranche, 1997. *Am J Med Genet* 77:31-37.

Steinmann B, Royce PM, Superti-Furga A (2002): The Ehlers-Danlos syndrome. In: *Connective Tissue and Its Heritable Disorders: Molecular, Genetic, and Medical Aspects* (P.M. Royce and B. Steinmann, eds.) 2nd ed, pp 431-523, Wiley-Liss, New York.

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