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

Patient information

Date: .....

NAME: .....

First name: .....

Date of birth: .....

City: .....

Informed consent:

I confirm that my clinical data presented here can also be used for medical research and scientific publications without my name (anonymous)

Signature: .....
(patient or parent/legal representative when applicable)

Place/date: .....

Motivation: .....

Referring physician: .....

Adress of referring physician: .....

.....

Referring department: .....

Phone: .....

Fax: .....

E-mail: .....

Material: *blood:* ❑ *DNA:* ❑ *fibroblasts:* ❑

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 proα1(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 O no O
- Arterial/intestinal/uterine fragility or rupture yes O no O
- Extensive bruising yes O no O
- Characteristic facial appearance (thin nose, small lips, starring eyes, hollow cheeks, missing ear lobe) yes O no O

Minor diagnostic criteria

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

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.

Version (August 2008) by:

Prof. emer. Dr. med. Beat Steinmann
beat.steinmann@kispi.uzh.ch