ADITUM Journal of Clinical Research and Clinical Case Reports

Open Access Mini Review

Ehlers-Danlos, Aneurysms and Arterial Dissections

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Article Info

Received: May 05, 2021
Accepted: May 23, 2021
Published: May 26, 2021

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Citation: Claude Hamonet, Denis Ducreux, Richard Amoretti. "Ehlers-Danlos, Aneurysms and Arterial Dissections". Clinical Research and Clinical Case Reports, 1(3); DOI: http;//doi.org/04.2021/1.1012.

Abstract

Aneurysms are part of the signs of Ehlers-Danlos' disease (or syndrome). They were initially described by Sack in 1932 and then by Barrabas in 1967. They were included in the "vascular forms" within the classifications established by consensus between geneticists, in Villefranche in 1997 and then in New York in 2017. An excessive dichotomy was established between the so-called serious "vascular forms" and the benign "non-vascular forms" often called hypermobile. For our cohort study, which we have been doing for 25 years and which includes 6200 patients who display typical clinical features of Ehlers-Danlos disease, it was impossible to separate the "vascular forms" from the "non-vascular" as all displayed hemorrhages. This disease is hereditary and systematically transmitted. We have in fact observed that all the children of which either the father or the mother was affected by the Ehlers-Danlos disease, were also affected (cohort of 62 families). We consider that all forms of Ehlers-Danlos can be complicated by aneurysms that must be systematically searched by MRI scanner.

Keywords: Ehlers-Danlos; arterial aneurysms; Ehlers-Danlos classification

Introduction

Aneurysms are part of the signs of Ehlers-Danlos' disease (or syndrome). They were initially described by Sack (1) in 1932, and then by Barrabas (2) in 1967. They were included in the "vascular forms" of the Ehlers-Danlos' disease within the classifications established by consensus between geneticists, in Villefranche (3) in 1997, Grahame (4) in 1998 and then in New York (5) in 2017.

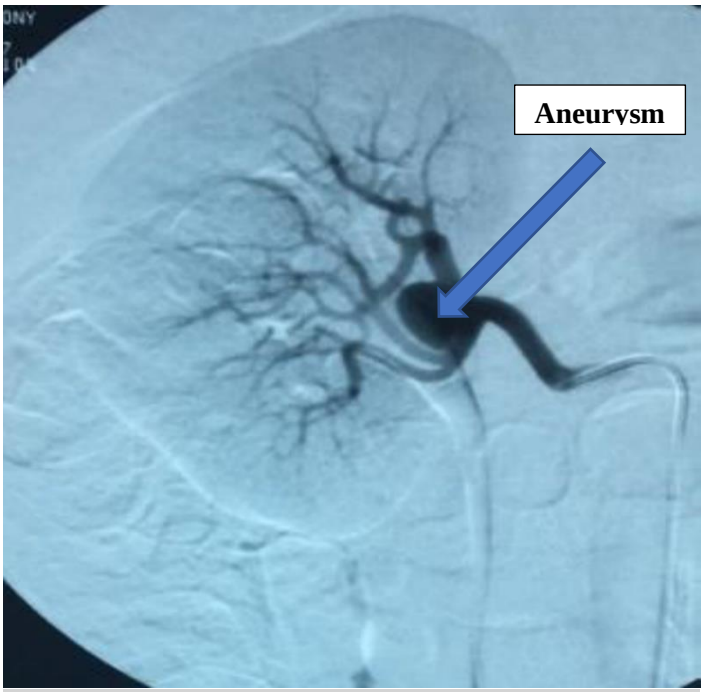
These forms, which have been opposed to the "non-vascular" forms of Ehlers-Danlos, considered to be less or even less severe, have a very bad reputation, with 50% of mortality rate before patients reach 48 years (5). In addition to aneurysmal ruptures and dissections, severe complications (hemorrhages, intestinal or uterine ruptures, peritonitis, intestinal occlusions) can occur. They are of great concern to patients who want to know whether they have "vascular form" or any of the 13 other forms identified (5).

Detecting arterial aneurysms in Ehlers-Danlos disease

Reading publications on Clinical identification of vascular forms with the exclusivity of aneurysmal risk is very difficult. All the signs identified and proposed, "facies of a madone" (6), a fine and transparent skin with a highly visible venous network, a tendency to bruises and hemorrhages, fatigue, frequent dislocation of the joints of the jaw, skin fragility, a precociously aged appearance of the skin of the hands, varicose dilation, early alopecia, are, in fact, very common in our active line of more than 6.200 patients without being associated with arterial aneurysm.

We therefore consider that there is no possibility of dissociating vascular and non-vascular forms by the clinical examination (7). Genetic tests (particularly COL3A1 research) often long or difficult to obtain and do not appear in practice as being an absolute test, especially if they are negative. Consequently this situation strongly suggests a systematic search for aneurysms by brain MRI of the aorta and its branches in any Ehlers-Danlos patient, especially if there is the suspicion of an aneurysm or a case of death suffered in a young subject among the relatives of the patient. Ultrasound is

insufficient, as we have seen with the discovery of several aneurysms (spleen, renal and femoral) in the MRI in a patient whose ultrasound, even repeated, could not reveal them. It is also possible to encounter an unusual location aneurysm, on a digital artery, as we have just discovered in a patient.



MRI scan of a renal artery aneurysm in a 58-year-old woman affected by the Ehlers-Danlos disease initially not diagnosed as "vascular"

Guidance

In the absence of an image of aneurysm, we suggest to renew the research every 5 years. In case of discovery of one or more aneurysms we recommend if their size is too voluminous a local surgical treatment taking into account the fragility of connective tissue of the Ehlers-Danlos syndrome. In case an aneurysm is detected, image monitoring shall be done at least annually. Obviously other treatments have proved their effectiveness and should be implemented: L-Dopa, oxygen therapy (every day), compression clothing and orthotics used every day and sometime during sleep, treatment of cardiovascular dysautonomy (POTS), treatment of stomach reflux and in particular constipation, treatments for dysuria or incontinence, treatments of menorrhagia, of endocrinal disorders (insipid diabetic), local and general treatments of pain (with xylocaine and TENS), treatments with cognitive (Ritalin) aim and sleep (melatonin).

Conclusions

Arterial aneurysms should be sought whenever a diagnosis of Ehlers-Danlos is made. The dichotomy: serious ("vascular") and benign ("non-vascular") forms (also called "hypermobile" or rather "hyperlaxis") must be abandoned. All forms of Ehlers-Danlos are potentially but rarely complicated by an aneurysm (around 2% of the cases according to an estimate based on observation of our cohort of 6200 cases). For every aneurysm patient a search for a hereditary connective tissue disease (the most frequent being Ehlers-Danlos) should be achieved together with any of its therapeutic consequences such as on dysproprioception, dystonia, pains, fatigue, cognitive, digestive and respiratory disorders.

This diagnosis leads to the detection of other family cases in this common disease which is transmitted to all children of the affected persons as we observe daily. This aspect is essential to thwart false accusations of violence on the part of parents (8) and to guarantee the benefit of appropriate medical follow-up and screening of possible complications among the children concerned.

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