

Ehlers-Danlos-Tschernogobow Syndrome: A Frequent, Rarely Diagnosed Disease whose Patients are often the Victim of an Abusive Psychiatrization

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Abstract

Despite its high frequency, Ehlers-Danlos disease remains practically unknown to doctors who evoke many other diagnoses with dangerous therapeutic and social consequences. These diagnoses primarily include mental disorders that sometimes lead to hospitalizations in psychiatry. Progression of the disease's clinical knowledge allows for a diagnosis of certainty, based solely on clinical examination, in the absence of biological marker in the hypermobile form, which is by far the most often encountered.

Keywords: Ehlers-Danlos hypermobile Type III; Iatrogenic; Hereditary disease; Cognitive impairment Anxiety

Introduction and Problem

Ehlers-Danlos disease was first described by dermatologists (Alexandre Nicolaïev Tschernogobov in Moscow in 1892 and Edvard Lauritz Ehlers in Copenhagen in 1900), and then it was confused with elastic pseudoxanthoma (PXE) by Alexandre Danlos [1-3]. The Russians gave him the name of its first descriptor. Secondly, Ehlers-Danlos Syndrome (EDS) emerged as the title of Achille Miget's thesis of Doctor of Medicine [4].

From the contribution of these authors, physicians have retained the skin's excessive stretch ability which remains one of the signs of EDS skin changes but it does not have "extraordinary elasticity", such as a "thin rubber blade" described by Danlos (Figure 1).

Articular hypermobility, already observed by Ehlers [2], has gradually become the key symptom of the disease, introducing under the denomination of benign articular hypermobility [5] the false idea, denounced by Rodney Grahame [6] that this clinical picture was rather another way of being normal than a pathological condition. As we have seen with our cohort of 2617 patients, the hypermobility present in childhood can diminish or disappear in adulthood. It can also be replaced by retractions from infancy [7], causing diagnosis errors. This hypermobility is usually measured by the Beighton test [8] which is often faulted and neglects the most hypermobile joint: The shoulder (Figures 1-3).

The publications of the last 20 years [9,10] have considerably enriched the clinical semiology, imposing a new description difficult to introduce, because of preconceived ideas and the multiplicity of symptoms that surprises doctors. However, this description is rapidly necessary for three reasons: risks of serious complications which can be prevented, the hereditary transmission which concerns all the children when one of the parents is affected, and the possibility of effective treatments which can considerably modify the quality of life of these patients [11].



Figure 1: Hypermobility of shoulders.

Clinical examination is the only way to arrive at the diagnosis due to the absence of genetic testing in the common, hypermobile systemic form which represents most of the cases encountered in mainstream medicine. The few modifications of collagen (Col 3A1 and COL 5A1 and COL5A2) identified, do not concern this form and are hardly usable in current medical practice. Six different types of EDS were described from this approach by collagen disorders [12] the contribution of cutaneous histopathology, very developed in Belgium [13], very probably represents a progress in the recognition of these patients most often rejected by doctors (Figure 4).

Suspect and Diagnose Ehlers-Danlos Disease

The diagnosis is based on the association of manifestations suggestive of changes in the connective tissue (frailty, alterations of biomechanical characteristics) on the one hand, and identification of other identical or similar cases in the family, on the other hand, signing the hereditary nature of the pathology.



Figure 2: Beighton test, Hypermobility of hips.



Figure 3: Excessive etirability of skin.



Figure 4: Eventration after abdominal surgery.

A recent study of 626 patients diagnosed with Ehlers-Danlos, confronted with the recognized descriptions [5,8] allowed us to retain a set of clinical manifestations enabling to make the diagnosis with certainty.

- Pains in multiple localizations (articular, abdominal, genital, migraines ...), difficult to relieve, evolving in crises on a continuous or sub-continuous background.
- Important fatigue on awakening, with feelings of body heaviness, "limbs in lead" and bouts of drowsiness.

- Motor disorders of proprioceptive origin with clumsiness, clash of obstacles, deviation of the walk, frequent sprains, dislocations or subluxations, involuntary dystonic contractions, falls.
- Hypermobility current or in childhood (to put one foot behind the head, side split). This does not exclude the presence of retractions (knees, ankles, feet or even elbows, from early childhood).
- Skin fragility with early stretch marks (before pregnancy) and / or very important, delayed healing, pathological scars, thin skin, transparent, soft, with sensations of electrical discharges in contact with metals, stretchability to a length at least equal to half of what the thumb and forefinger can grasp (Ehlers).
- Neurovegetative disorders (intolerance to vertical position, sweating, tachycardia, hypotension, hypothermia, chilliness, cold extremities, especially feet).
- Hemorrhages (bruising, gingivorrhagia, epistaxis, metrorrhagia).
- Hypersensitivity: Hearing, olfactory, vestibular, visual, cutaneous.
- Vision problems: Visual fatigue with blurred vision and image splitting.
- Respiratory blockages, shortness of breath on minimal exertion.
- Presence of cognitive and behavioral disorders: memory, attention, concentration, orientation, hyperactivity, anxiety.
- Other events may be associated. They are part of the clinical picture of EDS, contributing to strengthening the diagnosis: gastrointestinal (reflux, constipation, bloating), urological, sexual (dyspareunia), miscarriage, difficult childbirth, sleep disturbances, diffuse tonic or deficit crises, cysts and nodules in the skin or internal organs, pneumothorax, vascular complications (edema, lymphedema, arterial aneurysms (must be searched systematically by ultrasound or MR angiography), allergic reactions, sensitivity to infections (otitis), Vitiligo.

Diagnoses Confusion in Ehlers-Danlos Disease

Early diagnosis is even more necessary as these patients are very fragile and exposed to serious complications that can cause death. Fatalities may be due to the presence of aneurysms that are found in all forms of EDS and not only in those called "vascular". Otherwise, there are also iatrogenic complications (anticoagulant treatments, poorly

controlled surgical procedures, etc.). Among these iatrogenic effects, psychiatrization holds a very important place. Most of our patients face disbelief when describing their condition and doctors tend to be condescending when symptoms are enumerated. The use the phrase "it is all in the head" is far from uncommon. Doctors refer patients to psychologists and psychiatrists while blaming patients and the mother who, once in two cases, is also affected. This begins in childhood, with accusations of laziness, tendency to complain for nothing, insubordination and not staying still because of body perception disorders that force these kids to fidget constantly. This compromises schooling which is essential for the future of these children who, despite these difficulties, have excellent academic results. Adolescence usually marks, especially among girls, an accentuation of symptoms in a period, often difficult to live for them. The decisions of hospitalization and isolation from the family are not exceptional and have required repeatedly a direct intervention from the consulting physician. This is the case for one of our patients, then aged 15, who has since been a brilliant law student with responsibilities in a patient organization. This trend will continue into adulthood, leading to abusive diagnoses of depressive states, bipolar states and, of course, hypochondria, somatization, hysteria... The psychiatric therapies used (antidepressants especially) have very negative effects in these patients particularly reactive to the medications' side-effects.

The discourse of these patients can disorient the doctor during a first contact. They are readily voluble because they fear to forget what they must say to the doctor because of their memory problems and because of the very rich influx of ideas that disrupt their attention. On the other hand, one is struck by the relevance of the discourse, the ease, and the excellence of the reasoning. The performance of the academic results and the specialized tests confirm the very good level of intellectual performance of this population of patients. This advantage, combined with the usual dynamism and tenacity of these anxious patients, but most often non-depressed, allows them, within certain limits, to cope with the many difficulties encountered each day.

Conclusion

Ehlers-Danlos disease has been widely underestimated from epidemiological point of view (this is not a rare disease, it is common), severity (patients who suffer from it are fragile and often socially excluded) and transmission (it is systematic in all children of a person who is affected). This is therefore a very important universal public health problem in which awareness and appropriate measures can no longer wait.

Disclosure of Interest

The authors state that they have no conflict of interest.

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