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Ehlers-Danlos, an Unknown and Disturbing Syndrome. Free Comments

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Abstract

Ehlers-Danlos syndrome is a frequent, inherited disorder which is rarely diagnosed. This exposes these fragile patients to a long medical wandering and the risks of an iatrogenic that sometimes endangers their lives. It is a hereditary disease, without genetic testing in the most frequent clinical pictures that is transmitted to all the children of a parent who is affected. A definitive diagnosis is possible on a grouping of clinical signs. Cognitive disorders are common. The risk of aneurysms is possible due to arterial fragility. Cohabitation with autism and Asperser's disease exists and needs to be deepened.

Keywords: Ehlers-Danlos syndrome; Hyper mobility; Aneurysms; Proprioception disorders; bleeding; Asperser disease; Autism; Iatrogenic; Chronic pain; Cognitive disorders; Hereditary disease

Introduction

This frequent hereditary connective tissue disease has great difficulty in being admitted by the medical community. Its very existence is often questioned by some doctors who see in it the expression of a psycho-affective disorder. They thus deprive the concerned patients of diagnosis and adapted treatments. These patients are fragile and exposed to a long medical wandering and iatrogenic which can go as far as putting their lives in danger. We have diagnosed and often monitored and treated more than 5,000 people with this disease in 23 years. This intensive clinical experience has led us to many findings about this syndrome in medical practice, including neurological and psychiatric signs.

Identifying Ehlers-Danlos syndrome

It is possible today and we think that it is easy, for any knowledgeable clinician, on clinical symptoms alone. This is how two dermatologists: Tschernogobow, a Russian in Moscow and Ehlers [1,2] in Copenhagen, initially described it. They mentioned symptoms that have retained their strong relevance today: hemorrhages, joint instability and coetaneous fragility, motor control impairment (ataxic gait) wrote Ehlers. On the other hand, Danlos [3] was mistaken and described another illness: pseudoxanthoma elastic. He insisted on stretch ability and elasticity of the skin, which he describes as "extraordinary" [3]. This is the origin of many rebuttals in front of a skin, although a little stretchable, but not as stretchable as described by Danlos [3]. Other mistakes in the identification of this disease, polymorphic in its expression, will seriously compromise its identification, the most obvious error being that women complaining of pain, fatigue, sleeping and digestive disorders, precisely the characteristic of Ehlers-Danlos disease, were misdiagnosed with fibromyalgia instead of Ehlers-Danlos syndrome, which was unknown to the first descriptor [4].

The diffuse nature of the connective tissues involvement with damage to all human tissues is at the origin of the very large number of manifestations which confuse the clinician trained in semi logical analysis organ by organ. Moreover, this symptomatology is variable from one day to the next, posing, in the minds of doctors, the question of the organicity of the symptoms. The influence of internal factors (hormonal in the woman mainly), external (trauma, physical activity, climate variations) appears very clearly. Family sickness, it varies in expressions between members of the same family, including between heterozygous or homozygous twins as we have observed. Its evolution is impossible to foresee and one can, in the same family, observe severe forms with many disabilities and paucisymptomatic forms. In the absence of genetic testing for most Ehlers-Danlos syndrome's cases [5], it is possible to establish the definitive diagnosis on the basis of a grouping of clinical signs suggestive by their frequency and/or specificity. Twelve signs have been selected in a recent study [6] that overlaps the descriptions of other clinicians [7,8]. Clinical manifestations include arterial

aneurysms which are not the prerogative of the forms identified by geneticists [9] as "vascular". They may exist in all forms of the syndrome, especially in the brain, and must be systematically looked for, especially if the family has the notion of an aneurism or sudden death in a young subject.

3- Clinical criteria for the diagnosis of Ehlers-Danlos disease

The presence of five of the following 12 signs confirms the diagnosis of Ehlers-Danlos with a specificity of 99.10% and a sensitivity of 98.63% [6]: pains of multiple localizations evolving in attacks on a continuous background, very important fatigue; proprioceptive disorders of motor control with clumsiness, obstacle clashes, deviation from walking, falls; joint instability: sprains, dislocations/sublimations; thin, transparent skin that does not protect against electrostatic discharges, joint hyper mobility; retractions of knee flexor muscles in children; vasomotor disorders of the extremities (cold hands and feet), coetaneous and subcutaneous hemorrhages (ecchymosed); hyperacusis; constipation; gastro esophageal reflux disease. These clinical arguments are reinforced by the observation of identical family cases, which may be more or less obvious, proof of the hereditary nature of this connective tissue disease without genetic testing in the common forms.

4-Interpret a neurological or psychopathological symptomatology related to the Ehlers-Danlos syndrome Proprioceptive syndrome, omni present in Ehlers-Danlos syndrome, is a pathophysiological "key" for understanding and treating Ehlers-Danlos syndrome. The common feature in all these patients is the involvement of the connective tissue responsible for excessive frailty but also, excessive stretch ability, increased crushing potential and decreased elasticity. Sensitive and sensory receptors, subjected to mechanical variations of the connective tissue, send inappropriate information to the different systems of perception and adaptation of the body state. In this way, surprising demonstrations find their explanation. This is the case for neurovegetative disorders including changes in heart rate and blood pressure and postural orthostatic syndrome (POTS). This is the case of dystonia, which is very frequent [10] with sudden involuntary movements, intention tremor or postural retention, and sometimes generalized contractures. This is the case for many surprising neurological findings: hypoesthesia or localized anesthesia, non-systematized, loss sensations of a whole limb, a hemi corpus, with the sensation that it is not his body but that of another. These manifestations respond well to L-Dopa treatments. Elsewhere, they are pseudo paralysis of hemiplegic, paraplegic, quadriplegic or monoplegic topography, usually transient, but in rare cases, long lasting. These episodes, sometimes discrete in their neurological expression and rapidly transient, are often labeled with strokes with the risk of setting up an anti-platelet aggregation treatment that introduces a hemorrhagic risk. Pain of muscular origin in the lower limbs often lead to the diagnosis of sciatica or cruralgia, which is reinforced by the observation of the decrease in the height of an intervertebral disc, which is commonplace in Ehlers-Danlos syndrome made of generalized tissue flexibility. Cerebral imaging [11] highlights sub cortical lesions affecting white matter tracts which are interpreted as the consequence of shock waves on poorly shielded brains by pathological tissues which does not dampen them as well. Elsewhere, images of Leucoaraiosis of the supraventricular region by venous stasis may be confused with those seen in multiple sclerosis.

Cognitive difficulties are very often associated (neurology,%): memory disorders, attention, concentration, orientation in time and space, learning disabilities with dyslexia, dysorthographia, dyspraxia with negative consequences on schooling and working life. They usually contrast with observation, judgment and reasoning abilities that allow for compensation. The association with autism and Asperser's disease is manifested in two ways: by the behavior of certain children in a family with Ehlers-Danlos syndrome. They are hyperactive, gifted but having difficulties in contact with others. Elsewhere, it is the psychiatrists, trained in the diagnosis of the syndrome, who identify them.

Conclusion

Ehlers-Danlos syndrome is a clinical entity currently identifiable with certainty by the clinical inputs. The genetics contribution remains very modest in everyday practice. Advances in the clinic are sufficient to identify the disease, to inform about the risk of transmission to avoid certain dangerous therapies (anticoagulants and surgery) and to implement effective treatments (clothing with proprioceptive orthoses and oxygen therapy).

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