Journal of Arthritis
ISSN: 2167-7921

Hamonet and Brock, J Arthritis 2015, 4:2
DOI: 10.4172/2167-7921.1000148

Research Article Open Access

Joint Mobility and Ehlers-Danlos Syndrome, (EDS) New Data based on 232 Cases

Hamonet C* and Brock I

Department of Medicine, University Paris-East-Creteil (UPEC), 8 rue du general Sarrail, 94000, Creteil, France

Abstract

Hypermobility in Ehlers-Danlos syndrome was the first historical finding, and is still deemed by many to be a requirement for diagnosis. It was aptly described by Lauritz-Edvard Ehlers on December 15, 1900, at the Danish Society of Dermatology in Copenhagen. Hypermobility is also described in the clinical report of Henri-Alexandre Danlos and Achille Miget who, as the first, associated Ehlers and Danlos in the same syndrome. Currently, there is some controversy about the role of hypermobility, and on how it may be diagnosed. The discovery of muscle and tendon retraction as a part of this syndrome sheds new light on the diagnostic debate surrounding a genetic syndrome in the absence of biological markers, with as of yet no fully conclusive genetic test available for its identification.

Our study included 232 patients (84% women) from 2 to 70 years of age. Diagnosis was made in accordance with the Villefranche geneticists classification criteria. The patients enrolled in the study are of the hypermobile type.

We looked for muscle, tendon, and aponeurosis retraction in the knees, ankles, and the soles of feet. In our population of 232 patients, we found a retraction of the hamstrings in 203 individuals (87.5%). Retractions of the triceps surae were found in 90.9% of patients, and retractions of the soles of feet were observed in 95.9% of patients.

The impact of retraction on the Beighton palms-on-the-floor test is very great indeed 97.8% of patients who present a retraction of the hamstrings of over 45° cannot perform this maneuver. Hamstring retraction does not, however, affect the test of knee hypermobility (recurvatum).

The presence of muscle and tendon retractions in the posterior muscle compartments of the lower limbs and the soles of the feet constitute clinical features of Ehlers-Danlos syndrome. They should be addressed with a view to prevention and treatment, mainly through physical therapy.

Keywords: Ehlers-Danlos Syndrome; Hypermobility; Beighton Score; Contractures; Muscle retractions; Physical medicine

Introduction

Hypermobility was the first aspect of Ehlers-Danlos [1] to have been described. It was depicted by Lauritz-Edvard Ehlers, on December 15, 1900, in his presentation of the case of a 21-year-old law student to the Danish Society of Dermatology and Venereology in Copenhagen [2]. It was taken up again, in Paris, in 1908, by Henri-Alexandre Danlos [2,3], who emphasized a sign which has survived to this day, despite of it being frequently faint or absent: excessive skin elasticity. Danlos' description more closely resembles pseudoxanthoma elasticum rather than Ehlers-Danlos syndrome as it is described today.

In the presence of genetic tests that do as of date not cover all variants of EDS and given the large number of symptoms, which are highly subjective and variable, particular attention has in fact been given to the presence of hypermobility (or hyperlaxity) of the joints. The current trend is to rule out Ehlers-Danlos where hypermobility is absent. In clinical practice, the question is how to apply the criteria for pathological joint hypermobility. Beighton [4,5] chose a 9-point score to measure this sign, which has taken hold mainly among geneticists. In practice, this test is difficult to implement and to interpret, and was subjected to criticism [6,7] at the First International Symposium on Ehlers-Danlos Syndrome, at Ghent, Belgium, in September 2012. This is in accordance with the opinion of different authors [8,9].

Our intention was to contribute to this fundamental discussion on the identification of a pathology whose very existence continues to be questioned, exposing patients to one misdiagnosis after another, and subjecting them to iatrogenic accidents with most unfortunate consequences. We demonstrate (paradoxically, given the aforementioned context of hypermobility) the presence of muscle and tendon retractions which call into question the interpretation of the Beighton test, and deem it now necessary to discover other methods of analyzing this sign, which remains an important tool in the diagnosis of Ehlers-Danlos syndrome.

Patients and Methods

Our study examined a cohort of 232 patients, who were recruited at random, out of a clinic specializing in Ehlers-Danlos syndrome. In the absence of genetic testing, diagnosis was made on the basis of criteria arising from recent clinical trials [8,9] and which are compatible with the Villefranche geneticists' classification system [10]. The patients enrolled in the study were identifiable, in accordance with this classification as having the hypermobile form.

Four main criteria were selected:

- Cutaneous fragility, which is expressed by the thinness of the skin, through which the venous network is visible, early-onset or abundant

***Corresponding author:** Hamonet C, Ehlers-Danlos Clinic, Hospital-Hotel-Dieu de Paris, 1 place du Parvis Notre-Dame, 75181 Paris, Cedex 04, France, E-mail: pr.hamonet@wanadoo.fr

Received February 26, 2015; **Accepted** March 26, 2015; **Published** April 06, 2015

Citation: Hamonet C, Brock I (2015) Joint Mobility and Ehlers-Danlos Syndrome, (EDS) New Data based on 232 Cases. J Arthritis 4: 148. doi:10.4172/2167-7921.1000148

stretch marks, slow wound healing, and an excessive sensitivity to static electricity, with sensations of electric shock (sign of the car door opening), where the person feels an electric shock when he or she touches the car door handle).

- Joint hypermobility evaluated by a Beighton test equal to or exceeding 5/9 or having been hypermobile during childhood or adolescence (e.g., able to put one foot behind one's head, doing the splits, etc.).

- A joint or locomotor disorder manifesting as: pseudo-sprains; joint blockages or subluxations; clumsiness, especially when walking around, bumping into objects (the door sign,) in which the person bumps against the doorframe or catches on the door handle or knob.).

-The existence of at least one similar case in the family, a sign of the genetic nature of the condition.

To these main criteria, one may add other clinical manifestations [7-9] which also have great diagnostic value due to their frequency and their association: diffuse pain, fatigue, hemorrhages, dysautonomia, digestive problems (gastroesophageal reflux, constipation, bloating), respiratory problems (holds their breath, easily out of breath), bladder, sphincter and perineum problems, and also hyperacousia, hyperosmia, dizziness, teeth and mouth problems, dystonia, and cognitive disorders.

Study methods

We looked for muscle, tendon, and aponeurosis retraction in the knees, ankles, and the soles of the feet.

-For the hamstring muscles, we utilized the Lasègue maneuver (the patient is lying on his back, the examiner lifts the patient's heel with the ankle in a neutral position, knee straight, hips tight against the examining table, until the examiner encounters resistance which prevents the manoeuver from continuing. A goniometric measurement of the angle obtained between the plane of the examining table and the axis of the leg is taken. In the interest of simplifying the analysis of the data, the angle measurements were rounded to the closest of the following values: 10, 20, 30, 45, 60, 70, 80, 90, 100, measured in degrees. In general, muscle and tendon tension is not experienced as being painful; notwithstanding, one must be gentle so as to avoid subluxating the hips, which actually occurred with one patient. Retraction was deemed to exist when the angle between the patient's stretched-out leg and the examining table is equal to or less than 80 degrees.

-For the ankle, the patient is lying on his back, knees straight. The examiner moves the ankle by holding the heel in the palm of his hand and pushing with the front part of his forearm against the patient's forefoot. This uses all three heads of the triceps because the knee is extended, thereby putting tension on the head of the femur. A simple framework was established for evaluating the muscle and tendon retraction of the triceps: 1 denotes limited dorsal flexion of the foot, 2 signifies limited to 90 degrees, and 3 indicates it is impossible to achieve a right angle.

-For the soles of the feet, it is possible, during maneuvers which put the soles under tension by pressing on the forefoot and pushing backwards, to palpate rigid fibrous cords, which are sometimes painful, and are situated on the soles of the feet. These are an expression of a retraction either of the plantar aponeurosis or the flexor tendons of the toes. This can be measured by using a podoscope with the following scale, 1 if one-quarter of the sole is not in contact with the plate of the podoscope, 2 if half is not in contact, 3 if three-fourths of the sole are not in contact, and 4 if the entire sole of the foot is not supported in parallel, the Beighton score was calculated as follows, 1 point if the patient can touch the floor with the entire palm of both hands, with knees straight, with bare feet, heels on the floor; 2 points for passively touching the anterior forearm with the fleshy tip of the thumb on both sides; 2 points for being able to dorsiflex the 5th phalange at the metacarpophalangeal joint, with the wrist straight and fifth digit interphalange completely extended, to an angle of 90 degrees or above; 2 points for being able to passively hyperextend (recurvatum) both elbows by over 10 degrees; 2 points for being able to passively hyperextend (recurvatum) both knees by over 10 degrees with the patient lying on his back.

All measurements were made by the same doctor, who is a physical medicine & rehabilitation specialist.

Results

Description of the population studied 232 individuals aged 2 to 70 with an average age of females (84% of individuals) of 32 years (SD: 17.5) and of males (16% of individuals) of 20 years (SD: 14.1). Four age groups were defined for analysis: age 10 and under, 11 to 20 years old, 21 to 40, and 41 to 70.

Retractions and Ehlers-Danlos syndrome

Retractions of the hamstring muscles

Out of a population of 232 patients, a retraction of the hamstring muscles was found in 203 patients (87.5%).

Among the 203 retracted patients, the retraction is greater than or equal to 45 degrees in 41% of cases and between 10 and 44 degrees in the remaining cases.

Breakdown by age group is uneven

-In children aged 10 and under (32/38 patients): 84.2%.

-In the 11 to 20 year-old group (50/51 patients): 98%. In this group, half have retractions that are less than 45 degrees (Figure 1).

-In the 21 to 40 year old group (63/73 patients): 84.3%.

-In the 41 to 70 year old group (57/69 patients): 82.6%

These retractions can be found early on, in childhood.

The gender ratio changes with age among the retracted patients: 59.4% of female patients in the children under 10 group, rising to 69.6% in the 11-20 year old group (Figure 2).

A retraction of the triceps was found in 90.9% of patients. Retraction was severe (rating 2 and 3) in 50.7% of cases and moderate (rated 1) in 40.2% of cases (Figure 3).

Plantar retractions were found in 95.9% of cases. Retractions deemed to be moderate (rating 1 or 2) in 52% of cases and severe (rating 3 or 4) in 43.9% of cases.

Influence of retractions on the Beighton score

Impact of hamstring muscle retraction on the palms-on-the-floor test in the Beighton score (Figure 4).

In 98% of patients presenting this part of the Beighton test is impossible to perform (Figure 5)

In 73% of patients presenting moderate retraction, the test is impossible to perform.

In the group without retraction, the palms-on-the-floor test is positive in 75.9% of patients.

Figure 1: Retraction of hamstrings and sural triceps.

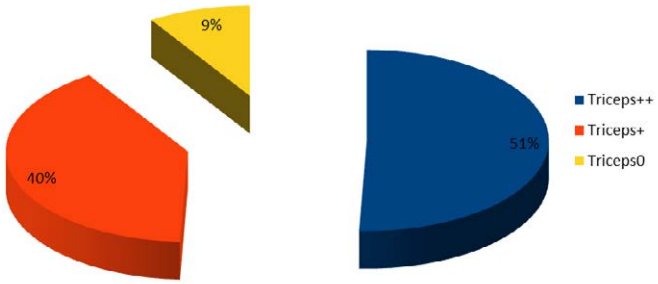

Figure 2: Retraction of the sural triceps.

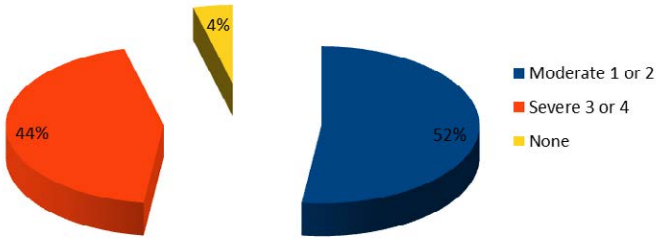

Figure 3: Plantar retractions.

Out of the total number of patients in this study, 177 patients cannot obtain this point on the Beighton score. Thus a retraction of the hamstring muscles deprives 84.2% of Ehlers-Danlos patients of one point on the Beighton score. The author of the Beighton score himself insisted on the fact that the palms-on-the-floor test involves the hips and not the spine. Therefore, a negative score on the test in the event of a muscle retraction which prevents the hips from flexing substantiates his statement.

Impact of hamstring muscle retraction on the genu recurvatum

29.3% of patients presenting a retraction greater than or equal to 45 degrees do not have genu recurvatum, neither do 28% of those who have an average retraction nor 20.5% of those who have a moderate retraction. Recurvatum is present in only 38.5% of the group without retraction.

Discussion

Muscle and tendon retractions and Beighton score calculation

In a patient who cannot touch the floor with both palms (one point out of 9), one must look for a retraction of the hamstring muscles. If this is present, then this part of the test must be excluded from the final calculation of the score since, in our series, almost all of the patients (96%) who were unable to perform in this test also had a retraction of the hamstring muscles. The presence of retractions of the hamstring muscles does not seem to have an impact on the genu recurvatum (two points out of 9 on the Beighton test). The percentage of people with recurvatum is no lower in this cohort than it is in the group without retraction.

Early-onset retraction of the hamstring muscles, the sural triceps and the plantar arch constitute a new sign in favor of a diagnosis of Ehlers-Danlos Syndrome

The quasi-omnipresence of retraction of the triceps (90.9% of patients) and of the soles of the feet (95.9% of patients) probably explains why children with Ehlers-Danlos syndrome tend to walk on the tips of their toes. Retraction contributes to difficulties in proprioception while standing up and walking, because of a poor perception of the characteristics of the ground. This is supported by the very significant improvements obtained by wearing appropriate orthopedic insole [11].

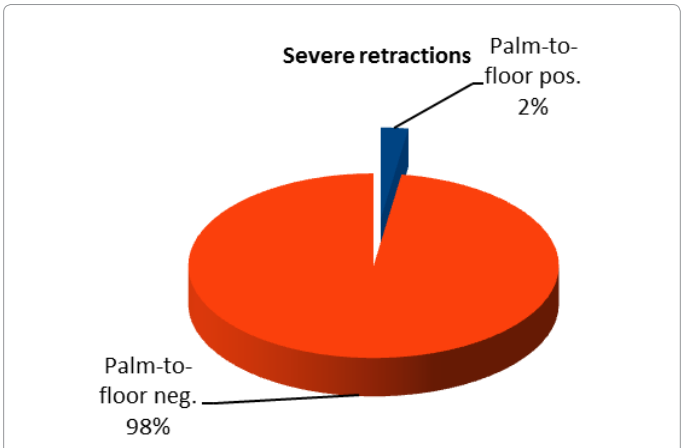

Figure 4: percentage of negativity of the palm-floor test with hamstring retractions greater than or equal to 45 degrees.

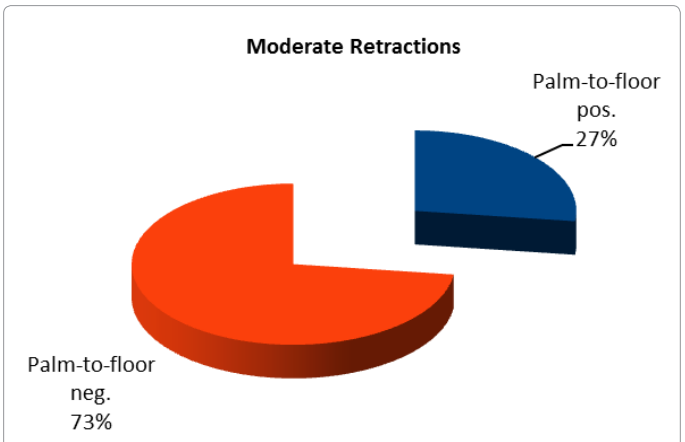

Figure 5: Palm-floor test fealure with moderate retraction of hamstrings.

The presence of a retraction of the hamstring and triceps muscles must not serve to rule out a diagnosis of Ehlers-Danlos syndrome; on the contrary, it should be seen as one of its characteristic signs, especially in children. Indeed, retractions are a hallmark of the clinical presentation of this syndrome. The distribution of retractions in the lower limbs is remarkable because they tend to be found mainly in the posterior muscle sheath. This can be objectively determined if the patient's heel can easily be made to touch the buttocks when lying face-down.

Retractions in the upper limbs are unusual, though they may be found in the elbows with an actual flexum of the elbow.

In addition to the Beighton score, what other means can one use to diagnose joint hypermobility?

This test has been criticized for its lack of precise measurements, the variability which exists from one observer to another, its susceptibility to errors, such as in cases where there are muscle and tendon retractions. It has even been suggested that the test be discontinued [4]. It is therefore important that improvements are made in the semiology of hypermobility in Ehlers-Danlos syndrome and how it is applied.

First, it must be noted that hypermobility varies with age in Ehlers-Danlos syndrome. There is a general tendency for hypermobility to diminish with age, although not in every patient. Diagnosis only requires that hypermobility was present at some point in the patient's life (for example, in childhood). One must remember, however, that it may be difficult to find evidence for hypermobility, because of the muscle and tendon pain that is so frequent in this syndrome.

A firm diagnosis of hypermobility can be made if the patient has ever in his or her life been able to put one foot behind his head or do the splits face forward.

The Beighton test fails to take the joints close to the arms into consideration. Not withstanding, the arms are very much affected by hypermobility: protruding shoulder blades, hypermobility, often accompanied by luxations of the scapulohumoral joint. Instability of the elbow, with hyperpronation is also a significant sign of hypermobility in Ehlers-Danlos syndrome.

Is multiple hypermobility an essential requirement for the diagnosis of Ehlers-Danlos syndrome?

No, we have encountered forms presenting without hypermobility but with all the other signs pointing towards this diagnosis and where other members of the family do present obvious signs of hypermobility. It remains, however, a highly conclusive sign, which is present in 97% of cases in a cohort of 644 patients [7]. In addition, the requirement of a Beighton score of 4 or 5, with no distinction made as to which joints are involved, seems quite arbitrary to us. The presence of highly pathological hypermobility in a single joint (for example, the scapulohumoral joint) may be sufficient to assert the existence of this sign. This is the direction underlying our work, and the reason why we propose two new measurements: the distance from heel to buttocks, with the patient lying face down, and the measurement of the glenohumeral abduction, while the shoulder blade is blocked (Figures 6 and 7).

What treatments should be offered in the presence of retractions in Ehlers-Danlos syndrome?

It seems appropriate to treat them with physical therapy, such as manual physical therapy postures and postural orthotics. Surgery would not seem to be indicated due to the risk of: hemorrhage (which is exacerbated in this pathology), healing problems, the risk of relapse, and the risk of algoneurodystrophy. Algoneurodystrophy is frequent in Ehlers-Danlos, including dysautonomia [7,9]. The injection of botulinum toxin is debatable, especially because dystonia is frequently associated with the disorder [9]. The best treatment is to provide regular prevention through passive mobilization of the joints in danger.

Conclusions

1. In Ehlers-Danlos syndrome, muscle and tendon retractions are frequently found in the posterior muscle compartments of the thighs and legs, in association with plantar retractions. They are typical of the clinical presentation of Ehlers-Danlos syndrome and therefore contribute to making the diagnosis, especially in children.

2. Their presence creates a bias in the calculation of the Beighton score, in addition to its other imperfections. Given the weaknesses of this score, diagnosis of hypermobility must be based on other clinical findings, in particular, glenohumeral mobility.

3. In addition to the combination of muscle and tendon retractions and hypermobility, there is also the question of Ehlers-Danlos syndrome presenting without hypermobility. The presence of other clinical signs of the syndrome and the existence of family members who are hypermobile are sufficient for a diagnosis to be made.

Figure 6: Test hill- buttock, contrasting with hamstring retraction.

Figure 7: Cypel test, Gleno-humeral abdcution over 90°.

4. Prevention and treatment of retractions, mainly via physical therapy, is indicated.

References

1. Danlos HA (1908) A case of cutis laxa tumors with chronic contusion elbows and knees, Bul.soc.franç of Dermatology and Syphilis.
2. Ehlers LE (1901) Cutis laxa tendency to hemorrhages in the skin, loosening several articulations (case for diagnosis). Dermat Ztschr 8: 173.
3. Miget A (1933) Ehlers -Danlos, Thesis for Doctor of Medicine, Faculty of Medicine of Paris .
4. Beighton P, Solomon L, Soskolne CL (1973) Articular mobility in an African population. Ann Rheum Dis 32: 413-418.
5. Beighton P, Horan F (1969) Orthopaedic aspects of the Ehlers-Danlos syndrome. J Bone Joint Surg Br 51: 444-453.
6. Ouelette G (2013) Minutes of the First International Symposium on the Ehlers-Danlos syndrome (EDS). Journal of Medical Rehabilitation 33: 3-6.
7. Hamonet Cl, Vlamynck E, Serre N (2012) "Ehlers-Danlos syndrome, retractions of muscles flexor of Knees and Beighton test".
8. Grahame R (2001) Time to take hypermobility seriously (in adults and children). Rheumatology (Oxford) 40: 485-487.
9. Bravo JF (2009) [Ehlers-Danlos syndrome, with special emphasis in the joint hypermobility syndrome]. Rev Med Chil 137: 1488-1497.
10. Beighton P, De Paepe A, Steinmann B, Tsipouras P, Wenstrup RJ (1998) Ehlers-Danlos syndromes: revised nosology, Villefranche, 1997. Ehlers-Danlos National Foundation (USA) and Ehlers-Danlos Support Group (UK). Am J Med Genet 77: 31-37.
11. Hamonet C, Vlamynck E (2012) Le pied dans le syndrome d'Ehlers-Danlos (SED) hypermobile. Apport des orthèses plantaires. Etude avec 100 personnes. Le Journal de l'orthopédie 13: 1975-1980.