



VIII CONGRESO NACIONAL DE ENFERMEDADES RARAS Simposium Internacional de Familiares y Afectados por Ehlers-Danlos

Mesa Redonda: *Últimos avances Internacionales sobre Ehlers-Danlos.* Modera: Pascual Doñate

UCAM Murcia 17 de Octubre de 2015

- **Diagnosis and treatments of Ehlers-Danlos-Tschernogubow syndrome.**
 - *A French experience on 2.340 patients over 17 years.*
 - Diagnostico y tratamiento del síndrome de Ehlers-Danlos-Tschernogubow en Francia. Experiencia de 17 años y 2340 pacientes.
 - **Hamonet C., Brock I., Jaussaud R., Bienvenu B., Metlaine A., Vlamynck E., Haïdar R., Gompel A., Baeza-Velasco C.**
 - Hôtel-Dieu de Paris, Universités de Paris (Créteil, Descartes), Caen, Nancy.
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Introduction, Born of a strightly clinical Process.

1996.

I receive, in my clinic, in Henri Mondor Hospital, a woman patient 43 years old. She feels very painful, she has difficulties to walk, suffers from a pelvic prolapse, bladder and anal incontinences and she is very tired. Physical therapy is very painful and inefficient. She has intense «*tonic*» crisis. She is deaf on her right side .

I found a joint hypermobility and a strechable skin. My Diagnosis: EDS with a rich symptomatology.

1997.

At the national French PRM congress, I show a poster of this patient. A PRM physician, also President of *AFSED* asked me: « *can you receive patients with EDS from our association* »? I said YES.



Internet as a tool of epidemiology

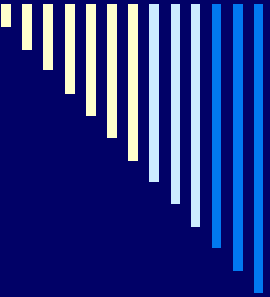
TWO years later, in 1998, I have seen 177 patients and made the following observations:

Late Diagnostic: in particular for women, diagnostic is achieved in average with a delay of 22 years

First description of disease based on observations of patients on my website for students

Result: A large amount of patients who discover the website quickly when searching the web with a couple of keywords and recognise their condition

Confirmation with a medical examination and follow up letter to the MD who then reports other patients



17 years later : 2340 patients Who are they?

(based on a sample of 279 patients examined between January, 1, 2014 and July 15, 2015).

Average age: 34 years (from 6 to 72 years).

Sex ratio: 2.77 (about 3 women to 1 man).

Average age of the first manifestation of symptoms: 9 years

Average age of diagnose completion: 31 years

Average diagnose delay: 22 years



ED Paris Scale

All the patients are examined with the same assessment tool.

18 Groups of clinical features: Pains, Fatigue, Sleeping problems, Joint disorders, Cutaneous fragility, Dysautonomia, Cardio-vascular signs, Hemorrhages, Deglutition, digestion & belly, State of bladder & urinary sphincter, Bucco/dental manifestations, ENT manifestations, Ophthalmological manifestations, Respiratory manifestations, Genital functions, Cognitive functions.

79 items

5 levels of severity (0, 1, 2, 3, 4)



Validity of Ehlers-Danlos Paris scale (EDSPS)

- A correlation study between observers has given positive conclusion
 - A comparative study between EDS patients and non EDS patients was conclusive in confirming the efficiency of EDSPS
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Results: signs present more 90%

- Hypermobility (97%)**
 - Multiple joint pains (95%)**
 - Fatigue (93%)**
 - Thin skin (94%)**
 - Genital Hémorrhages (90%)**
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A new hypermobility test: Cypel test. More 90 degrees abduction of gleno-humeral joint



Heel-bottom test in contrast with the hamstrings contracture





The clinical features present more 80% and less than 90%

- ❑ Awkwardness, motor control disorders (87%)
 - ❑ Fréquent ecchymosis (84%)
 - ❑ Cold feets (82%)
 - ❑ Muscles pains (87%)
 - ❑ Plantar contractures (82%)
 - ❑ Migraines (82%)
 - ❑ Visual fatigue (82%)
 - ❑ Sleeping problems (87%)
 - ❑ Luxations/subluxations (82%)
-



Fréquent ecchymosis (84%)



Plantar contracture (80%) = « *ED-foot* »





Luxations/subluxations (82%)





Signs are present more 70% & less than 80%

- Sprains or pseudo sprains (74%)
 - Hyperacusis (76%)
 - Reflux (72%)
 - Attention (72%)
 - Dyspnea (79%)
 - Recurrent URI (74%)
 - Excessive skin stretching (77%)
 - Difficult scarring (71%)
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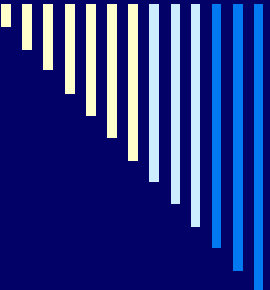
Excessive skin stretching (77%)





Difficult scarring (71%)





The clinical features between 60% & 70%

- Acute respiratory crisis (64%)**
 - Cutaneous hyperesthesia (68%)**
 - Hamstrings contractures (68%)**
 - Dystonia (64%)**
-



Hamstring contracture: impossible to put the palms of hands on the ground



Correlations (S. & M. Pommeret) of severity between groups of different items (422 patients):

1/ (left) skin/dysautonomy,
2/ (right) proprioception/sensorial perceptions.

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Date : mardi 15 septembre 2015

Corrélation 1-4

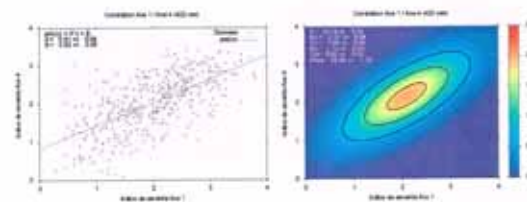
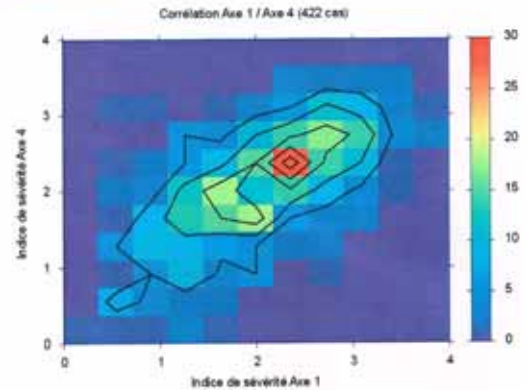


Figure 21 : Corrélation entre les valeurs de l'axe 1 et de l'axe 4.

Figure du haut : Données brutes avec les indices de sévérité divisés pour chacun des axes en 11 segments, la couleur indique le nombre de cas de 0 (bleu) à 30 (rouge).

Figure en bas à gauche : Chaque point représente un cas et la ligne droite représente la régression linéaire.

Figure en bas à droite : Ajustement paramétrique de la surface de corrélation par une distribution normale bidimensionnelle.

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Corrélation 3-5

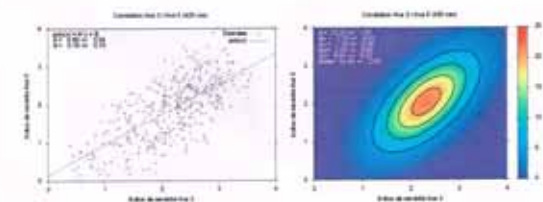
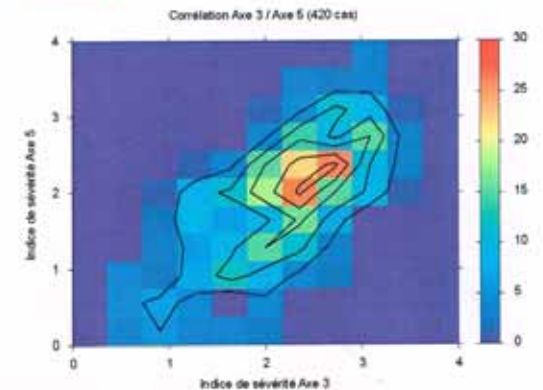


Figure 25 : Corrélation entre les valeurs de l'axe 3 et de l'axe 5.

Figure du haut : Données brutes avec les indices de sévérité divisés pour chacun des axes en 11 segments, la couleur indique le nombre de cas de 0 (bleu) à 30 (rouge).

Figure en bas à gauche : Chaque point représente un cas et la ligne droite représente la régression linéaire.

Figure en bas à droite : Ajustement paramétrique de la surface de corrélation par une distribution normale bidimensionnelle.



The clinical diagnose of EDS is possible through a concentration of very self explanatory signs

- **Widespread lesions** (systemic) typical of a damage to the conjunctive tissue. In particular signs gathering a skin fragility, proprioceptive troubles, dysautonomia and sensorial alterations (sight, hearing, smell, walking troubles).
 - **Presence of these signs in the patient family** (even if discrete), evidence of an autosomic atypical transmission because all the children with a parent affected by the disease are themselves affected.
 - Because **treatments improve** the functional state.
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The TREATMENTS

First International Francophone Symposium

"Treatments for Ehlers-Danlos Syndrome (EDS)"

*Medical School of Paris-East University Creteil, March
7th 2015*



***We all have obstacles to overcome to make a
difference and increase EDS awareness***

Against proprioceptive Disorders (limbs, trunk) : shape memory cushions, physical therapy, sports.



Rigid trunk orthosis



Hand and foot orthosis



Upper limbs orthosis: hand rest splint, shoulder scarf of Montreal



Compressive garments



Lower limbs orthosis (exoskeletons)



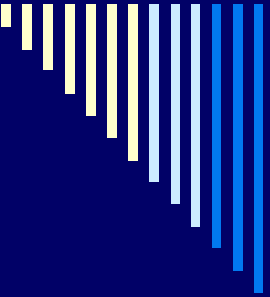
Molds seats at home & at school





Against proprioceptive disorders

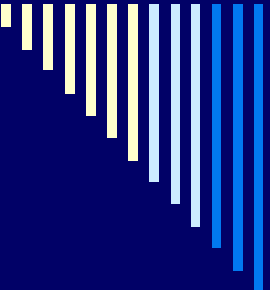
- Physical therapy, occupational therapy, Physical activities, sports (riding, Tai chi Chuan, swimming), balneotherapy, thermalism, using of orthosis.
 - Relaxation, hypnosis,
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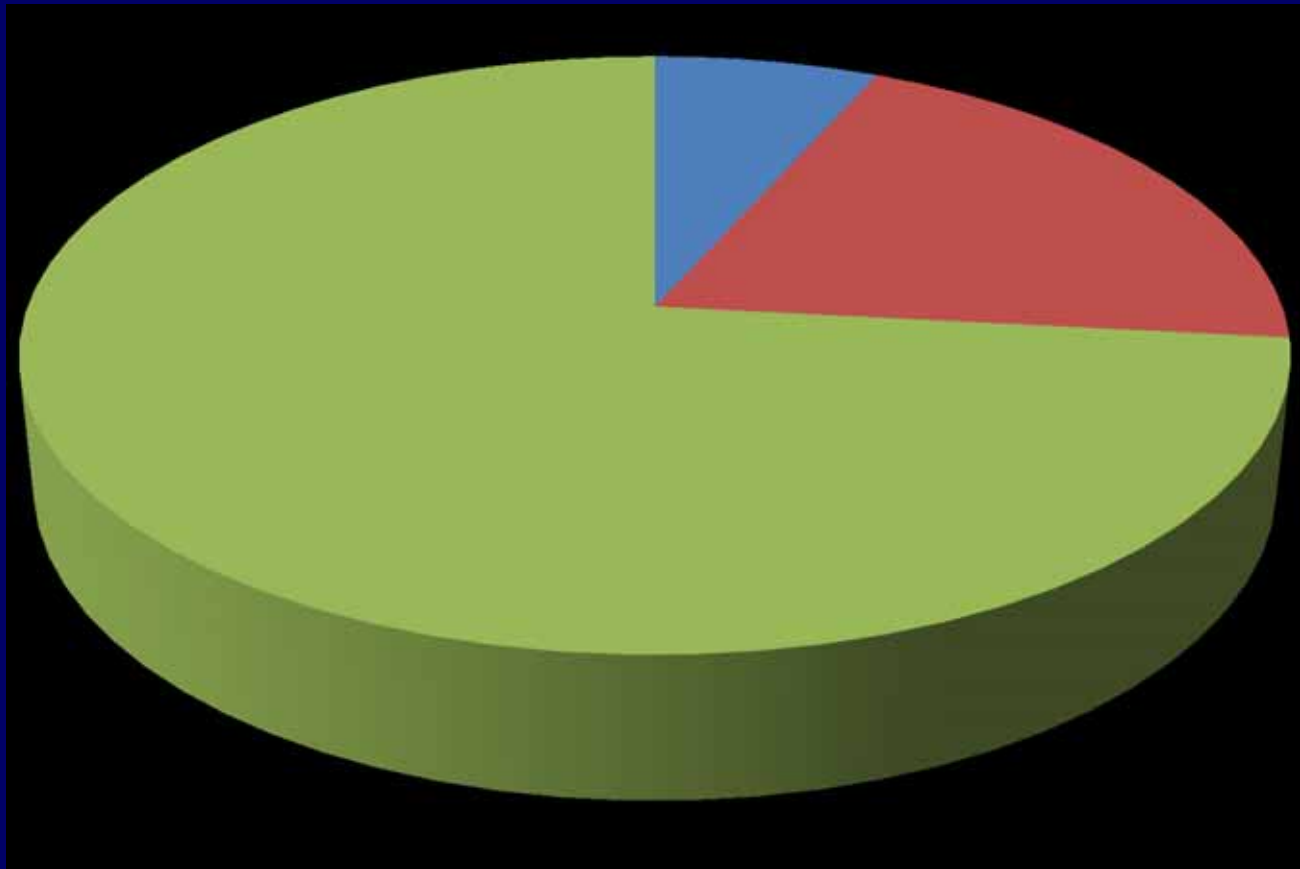
Treatment of respiratory proprioception disorders, fatigue, migraines...

Oxygenotherapy Percussionnaire



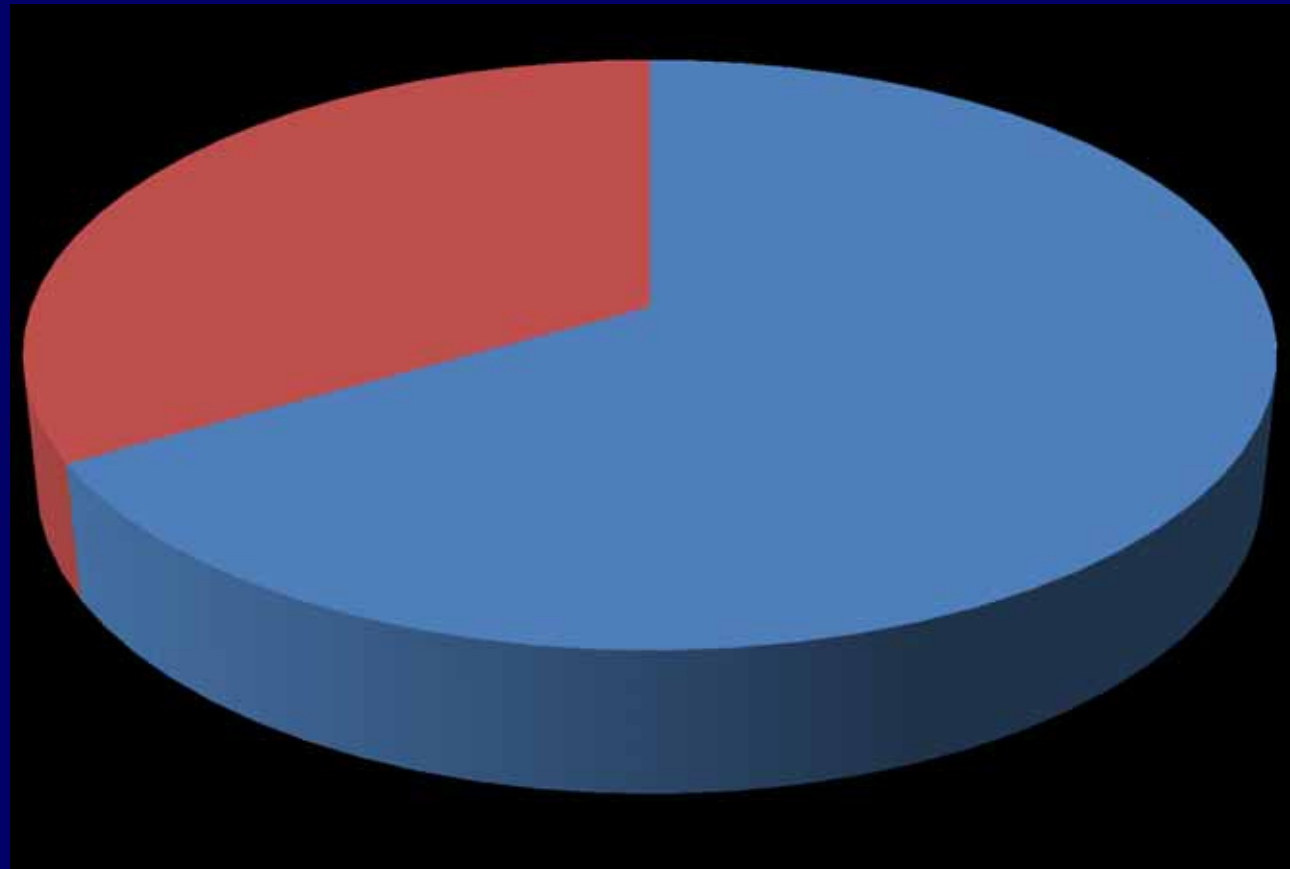


Action de l'Oxygène sur la fatigue après un mois de traitement (vert très bon, rose bon, bleu pas d'effet)





Effets de l'oxygénothérapie après un mois sur les migraines: Bleu, amélioration importante, rose amélioration partielle.





Against pains

Local treatments are preferred

TENS, Lidocaïne (patches, local injections) Heat, Massages, K-taps. **No** osteopathic manipulation.

General treatments must be selected: Baclofène, **L-Carnitine**, paracetamol+codéine tramadol, **nefopam**. Psychological training of pain.

No morphine, **no** cortison.



Others

Antidystonics : L-DOPA

Beta blockers (Bisoprolol 1,25 à 2,50/j.)

Therapy against gastro-duodenal reflux,
constipation, bladder disorders...

VitamineD, vitamine C

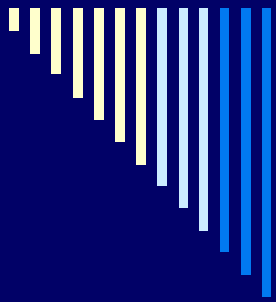
Neuropsychological and speech therapy
rehabilitation (attention, memory)

Psychological support for the patient and his
family.



Conclusion générale

- Ehlers-Danlos disease now **is not a Rare disease.**
 - Its diagnosis is easy on **clinical aspect and family character** without genetic test and cutaneous biopsy.
 - Le **historical difference** between several forms must be **revised**: all the persons with an EDS can have an arterial aneurism (« ***all the EDS are Vascular*** »). It will be researched and treated if necessary.
 - The dominant idea tht only the EDS-vasc are severe and that « anothers » (« hypermobiles ») have not problems is FALSE and will be fight: **all the patient with an EDS can be in severe disabling situation**
 - **The EDS is the first causis of Fibromyalgies** and this diagnose will be discussed sytematically.
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**Thank you very much for your friendly
attention**

Merci beaucoup pour votre invitation et de
m'avoir donné le plaisir de cette
première rencontre.

Muchas gracias
