Diagnosis and treatments of Ehlers-Danlos-Tschernogubow syndrome.

A French experience on 2,340 patients over 17 years.


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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 stretchable 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.


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
Results: signs present more 90%

- Hypermobility (97%)
- Multiple joint pains (95%)
- Fatigue (93%)
- Thin skin (94%)
- Genital Hémorrhages (90%)
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 feet (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%)
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.
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.
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 orthesis.
- Relaxation, hypnosis,
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.
Antidystonics: L-DOPA
Beta blockers (Bisoprolol 1.25 to 2.50/j.)
Therapy against gastro-duodenal reflux, constipation, bladder disorders...
Vitamin D, vitamin 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 that 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 systematically.
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*