

2017: Ehlers-Danlos

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- In March 2017, EDS researchers around the world published new standards and guidelines for EDS and related disorders in the American Journal of Medical Genetics

Our time is now.

Ehlers-Danlos Syndromes
2017 International Classification

Supplement to American Journal of Medical Genetics
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Seminars in Medical Genetics

The Ehlers-Danlos Syndromes: Reports from the International Consortium
on the Ehlers-Danlos Syndromes
Guest Editors: Brad T. Tinkle, Fransiska Malfait, Clair A. Francomano and Peter H. Byers

- First mentioned by Hippocrates 4th century BCE
- Elastic Skin Man
- India Rubber Man
- Human Pretzel
- 1930 “Ehlers-Danlos”

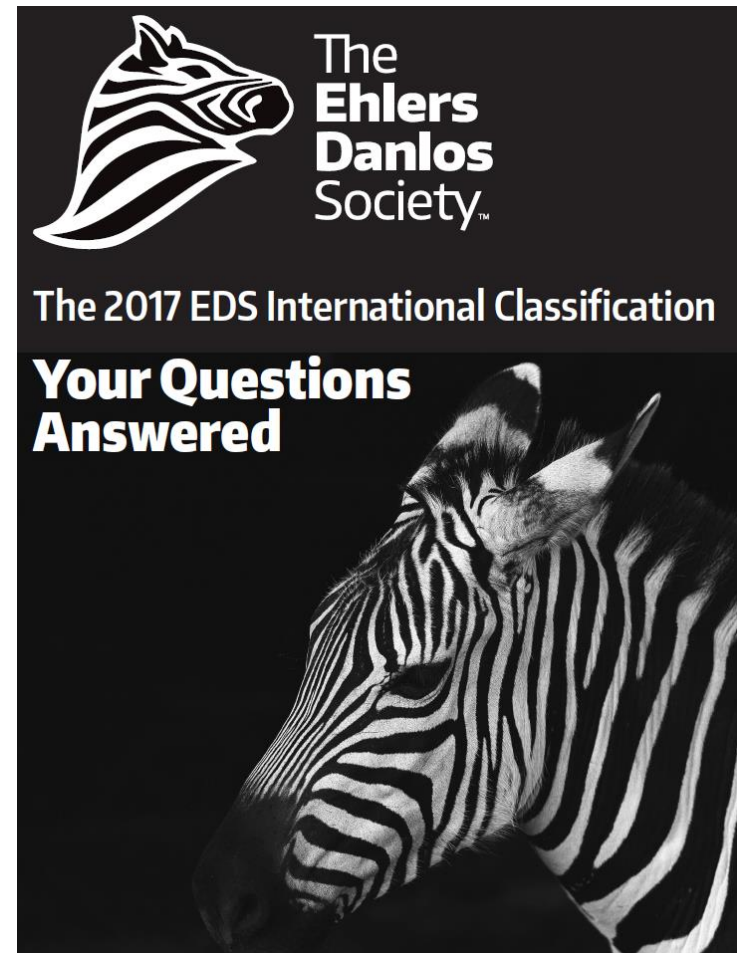


But Why???



- 1986 Berlin Meeting
 - 11 subtypes
 - Roman numbers
- 1998 Villefranche
 - 6 subtypes
 - Descriptions
- 2012 Ghent
 - Research/Registries
 - Keep up with changes

- Changes in names
- Need genetic testing to confirm any diagnosis other than hEDS
- Hypermobility EDS now either hEDS or Joint Hypermobility Spectrum Disorder



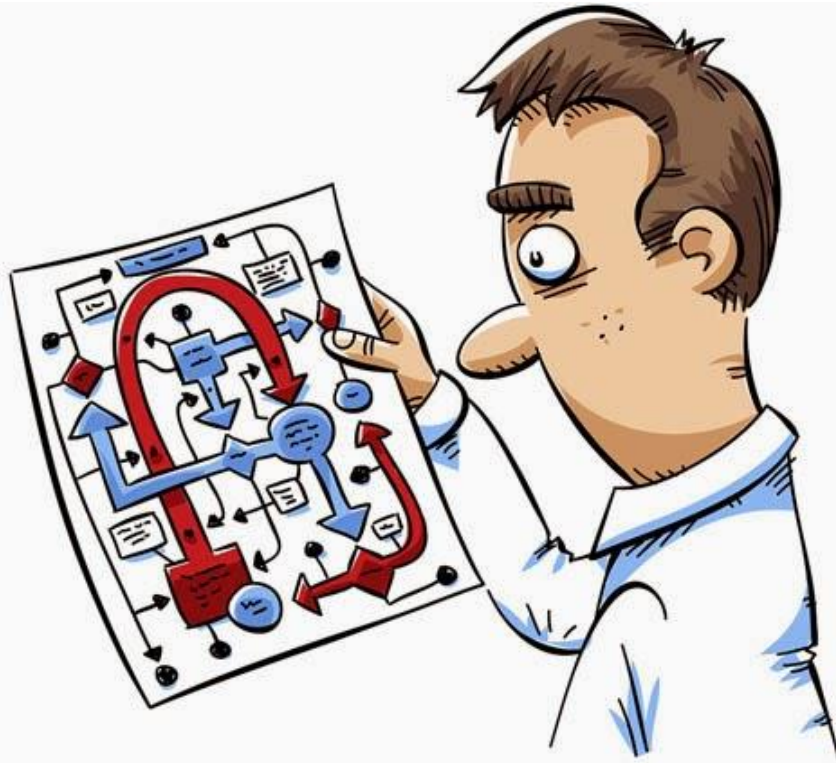


- Classical
 - cEDS Col1A1
- Classical-like
 - clEDS Tenascin-x
- Cardiovalvular
 - cvEDS COL1A2
- Arthrochalasia EDS
 - aEDS COL1A1/COL1A2
- Dermatosparaxis
 - dEDS ADAMTS2
- Kyphoscoliotic
 - K EDS PLOD1/FKBP14
- Brittle cornea syndrome
 - BCS SNF469/PRDM5
- Spondylodysplastic
 - spEDS B4GALT7
 - spEDS B3GALT6
 - spEDS SLC39A13
- Musculocontractural
 - mcEDS
D4ST1/CHST14/DSE
- Myopathic
 - mEDS COL12A1
- Periodontal
 - pEDS COL3A1/C1R/C1S

NOT TO PANIC!

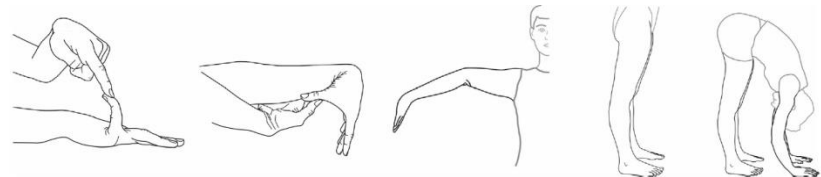
- Many people with EDS hypermobility form would not currently meet criteria for hEDS
- If you have hypermobility EDS you do NOT have to be re-diagnosed!
- hEDS is NOT better or worse than “Joint Hypermobility Spectrum Disorder”
- Problems and treatments are the same
- May have different diagnoses in same family





- Have to meet 3 criterion
 - Generalized joint hypermobility
 - Systemic problems
 - Must have at least 2 of 3
 - Must not have evidence of another connective tissue problem
- Go to EDS website!

- Generalized joint hypermobility
 - ≥ 6 preteen
 - ≥ 5 Teens to 49
 - ≥ 4 older than 50
- History 1 extra point
 - Can you know or could you ever touch both palms to floor
 - Ever touch thumb to forearm
 - Do Splits or joint tricks
 - Dislocate knee or shoulder more than once as a child or teen
 - Do you consider yourself double jointed



- Must have at least 2 of criterion 2 A, B and C
- 5 of 12 of feature A



A Systemic

1. soft skin
2. mild skin stretchiness
3. odd stretch marks
4. piezogenic papules both heels
5. recurrent or several hernias
6. atrophic scarring at least 2 places
7. pelvic floor prolapse without a pregnancy
8. Dental crowding or high palate
9. arachnodactyly
10. armspan/ht ≥ 1.05
11. \geq mild mitral prolapse
12. aortic root dilation $\geq +2$



- B Family History
 - 1st degree relatives
 - Parent, child, sib
 - Under **new** diagnostic criteria

Criterion 2

Systemic, Family History & MUSCULOSKELETAL



University of Iowa
Stead Family
Children's Hospital

- Pain in 2 or more limbs at least 3 months
- Chronic widespread pain at least 3 months
- Recurrent dislocation or major instability
 - ≥ 3 dislocations same joint
 - Or ≥ 2 dislocations different joints
 - Medical confirmation joint instability

- Must have at least 1



Criterion 3

“NOT THIS...”

All must be met

- Unusual skin fragility
 - Signs of other EDS
- Other medical problem
 - Rheum
 - Marfan
- Other joint/NM problem
 - Low muscle tone
 - Neuromuscular
 - Osteogenesis imperfecta



- The changes are basically in words
- One diagnosis is not better or worse than the other
- You do not need to be Re-diagnosed
- Don't need to do echoes as often for hEDS
 - One normal as child and one as adult
- Best treatment
 - Exercise
 - Drink more fluids
 - Eat more salt
 - Cognitive therapy



