EDNF 2015

EDS – PAST, PRESENT & FUTURE

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EDS

AN INAUSPICIOUS PAST
EDS

THREE UNFORTUNATE ERRORS!
FREDERICK PARKES-WEBER (1863-1962)
1200 medical articles + 23 books over 50 years; 7 eponymous diseases including Osler-Weber-Rendu disease

SUGGESTED THE NAME EHLERS-DANLOS SYNDROME IN 1936
Edvard Ehlers (1863-1937) Denmark

1899 Paris Society of Syphilology and Dermatology.

The patient he presented was a 21 years old late walking; frequent subluxations the knees; haematomata on minor trauma; extensible skin and lax digits.

1908 Henri-Alexandre Danlos 1844-1912 Paris gave a presentation to the same Society

Boy with skin extensibility/fragility of the patient's; 'pseudo tumors' in a patient with an inherent defect which he termed 'cutis laxa'.
A.N. CHERNOGUBOV [1892] Proceedings Moscow Venereology & Dermatology Soc - A case of cutis laxa:

“Intelligent 17-year-old peasant, starting walking aged 2+; falls; dislocated hip and elbow; thin, velvety, fragile, hyperextensible skin with normal recoil; painful joints with “extreme mobility far beyond the normal range”.

Denko CW J Rheumatol 1978;5(3):347-52. Chernogubov's syndrome: a translation of the first modern case report of the Ehlers-Danlos syndrome. This work is justifiably recognized as a seminal publication in rheumatology!
EDS III v HMS

THE FORTY YEAR SAGA!

WHEN RHEUMATOLOGISTS AND GENETICISTS DIDN’T TALK TO ONE ANOTHER (1 NOTABLE EXCEPTION)!
HMS 1967 (KIRK et al)
RHEUMATOLOGY

ERIC BYWATERS

EDS III 1968 (BEIGHTON)
GENETICS

VICTOR McKUSICK
"THE HYPERMOBILITY SYNDROME"

3

Musculoskeletal symptoms in the presence of generalised joint laxity in otherwise normal subjects".
“Another view is that isolated ligamentous laxity is a mild mesenchymal developmental disorder which lies at one end of a spectrum of heredofamilial connective tissue disease with the fully-developed picture of MFS or EDS at the other [Brown, Rowatt & Rose 1966]. This view was rejected with disastrous consequences!
HMS 1967
(KIRK et al)
RHEUMATOLOGISTS
JOINTS
OVERLAP WITH HDCTs
BRIGHTON (1998)
ANXIETY & PHOBIAS
(Bulbena 1988-)
AUTONOMIC DYSFUNCTION
(Gazit 2003)
GASTROINTESTINAL DYSMOTILITY
(Zarate 2010)

EDS III 1968
(BEIGHTON)
GENETICISTS
GENETICS
HDCTs
BERLIN (1986)
VILLEFRANCHE (1997)
CHRONIC PAIN (Sacheti 1997)
AUTONOMIC DYSFUNCTION
(Rowe 1999)
GASTROINTESTINAL DISORDERS
(Levy et al 1999)

TINKLE et al 2009
‘INDISTINGUISHABLE FROM ONE ANOTHER’
THE 9-POINT BEIGHTON HYPERMOBILITY SCALE
PHYSICAL PROPERTIES OF THE SKIN IN THE
EHLERS-DANLOS SYNDROME*

BY

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From the Clinical Research Division, Kennedy Institute of Rheumatology, London, W.6, and
St. Thomas's Hospital, London
‘THE RUBBER GLOVE SIGN’
MARFAN
AORTIC DILATATION
ECTOPIA LENTIS

MARFANOID
HABITUS

STRETCHY SKIN:
PAPYACEOUS SCARS:
STRIAE ATROPHICAE

JOINT HYPERMOBILITY
SYNDROME (=EDS III?)

OSTEOPENIA

OSTEOPOROSIS;
FRACTURES

OSTEOGENESIS
IMPERFECTA

EHLERS-DANLOS
CLASSICAL,
VASCULAR, ETC
TYPES
JOINT HYPERMOBILITY SYNDROME [JHS] IS A BENIGN, MULTI-SYSTEM, HERITABLE DISORDER OF CONNECTIVE TISSUE WITH OVERLAP FEATURES
Revised “1998 Brighton” diagnostic criteria for the Benign Joint Hypermobility Syndrome

<table>
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<tr>
<th>MAJOR CRITERIA</th>
<th>MINOR CRITERIA</th>
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<tbody>
<tr>
<td>▪ Beighton score ≥ 4/9 or (currently/historically)</td>
<td>▪ Beighton score of 1, 2, 3/9 (0, if aged 50+)</td>
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<tr>
<td>▪ Arthralgia &gt; 3 months in ≥4 joints</td>
<td>▪ Arthralgia in 1-3 joints/ back pain/spondylosis/spondylolysis/olisthesis.</td>
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<td>▪ Dislocation in &gt;1 joint, or in 1 joint on &gt;1 x</td>
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<td>▪ ≥ 3 soft tissue lesions</td>
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<td>▪ Marfanoid habitus</td>
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<td></td>
<td>▪ Skin: striae, thin, stretchy, abnormal scarring.</td>
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<td>▪ Eye signs: drooping eyelids or myopia</td>
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<td>▪ Varicose veins/hernia/uterine/rectal prolapse</td>
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The BJHS is diagnosed with:

- 2 major criteria or
- 1 major and 2 minor criteria or
- 4 minor criteria.

2 minor + 1° degree relative.

BJHS is excluded by presence of Marfan or Ehlers-Danlos syndromes (other than the EDS Hypermobility type formerly EDS III) as defined by the Ghent 1996 and Villefranche 1998 criteria respectively.
EDS

A BEWILDERING PRESENT
MUSCULOSKELETAL PAIN/JOINT INSTABILITY
OVERLAP WITH HDCT/SKIN/HABITUS
UTERINE/RECTAL PROLAPSE
CHRONIC PAIN SYNDROME
ANXIETY/PHOBIAS
DYSAUTONOMIAS
GI DYSMOTILITY
PROGRESSIVE DISABILITY
MUSCULOSKELETAL PAIN/Joint Instability
Overlap with HDCT/Skin/Habitus
Uterine/Rectal Prolapse
Chronic Pain Syndrome
Dysautonomias
GI Dysmotility
Progressive Disability
1967
1970
1980
1990
2000
2010
2015
Anxiety/Phobias
Chiarl + CranioCervical Instability + Tethered Cord
Mast Cell Activation
THE HYPERMOBILITY UNIT

ADULT RHEUM

PAED RHEUM
PAEDS GENERAL

CLINICAL GENETICS

RESARCH TRAINING

GASTRO

AUTONOMIC CV-MED

PHYSIOTHERAPY
OCCUPATIONAL THERAPY
PODIATRY
PAIN PSYCHOLOGY
ADMIN

ORTHOPAEDICS
OBSTETRICS
PLASTIC SURG
CARDIOLOGY
NEUROLOGY
NEUROLOGY
SPORTS MED
PAIN MEDICINE
ENDOCRINE etc.
A HOPEFUL FUTURE!?
THE DARK SIDE

- Genetic research yield so far disappointing
- Still don’t have a genetic marker for HEDS
- Genetic engineering is decades away!
- Breakthroughs do happen!
- People with EDS badly let down by the medical profession
- Patients are not listened to, believed, diagnosed or correctly treated
- Tragedies occur!
- I know of no other disease where this is so.
WHY THE NEGLECT/DENIAL?

- Concede that EDS has been a moving target
- Neglect of traditional clinical skills
- Over-reliance on laboratory findings
- Myths from the past – ‘normal subjects’
- Rheumatologists not taught about EDS
- Ideas about H/Mobility are those of 1960s
- Unaware/denial of PoTS/GI/Pain/Mast cell
- Incapable of making a definitive diagnosis
- Misdiagnosis: FMS, CFS, IBS, FII
- Failure to listen to what patients tell them
- Assumption that ‘it is all in the mind’
THE BRIGHT SIDE

- BENEFICIAL IMPACT OF ADVANCES IN MEDICAL SCIENCE ON OUR UNDERSTANDING OF EDS
  - BASIC SCIENCES
  - EPIDEMIOLOGY
  - MEDICINE
  - SURGERY
  - IMAGING
  - PSYCHOLOGY
  - PHYSICAL THERAPIES
  - ETC, ETC.
THE BRIGHT SIDE

- GROWING BAND OF DEDICATED HEALTHCARE PROFESSIONALS WORKING IN FIELD OF EDS
- WE NOW HAVE TREATMENTS THAT WORK
- EDS NATIONAL DIAGNOSTIC CENTRE [UK]
- EDNF CENTER FOR RESEARCH & CLINICAL CARE [US]
- TREATMENT CENTRES OPENING UP- UCLH RNOH; HMU [HJE]. BARTS HEALTH, COPE
- VIBRANT SELF-HELP GROUPS: EDNF, EDS UK, HMSA etc
- FORMATION OF EDS INTERNATIONAL
WE NEED:

- MORE EDUCATION FOR HEALTH PROFESSIONALS
- MORE TREATMENT FACILITIES TO CATER FOR NATION’S NEEDS
- MORE FINANCIAL RESOURCES FOR:
  - PATIENT CARE
  - EDUCATION
  - BASIC, TRANSITIONAL, CLINICAL AND EPIDEMIOLOGICAL RESEARCH
- MORE ASSERTIVENESS
- MORE CAMPAIGNING - ‘PEOPLE POWER’
No other disease in the history of modern medicine has been neglected in such a way!

Not just a USA problem; it is a world-wide problem!

It is a topsy-turvy world where patients often know more about their condition than their doctors! Anon!
GK Chesterton 1874-1936

"The Point of a Pin" in *The Scandal of Father Brown* (1935)
It isn't that they can't see the solution.

It is that they can't see the problem.

GK Chesterton 1874-1936

"The Point of a Pin" in *The Scandal of Father Brown* (1935)