Ehlers-Danlos Syndrome
Hypermobility Type (hEDS)

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No Disclosures

Objectives:
1) Review diagnostic criteria of the hypermobile form of Ehlers-Danlos syndrome
2) Review the major disorders of connective tissue in the differential diagnosis
3) Identify the basic treatment of EDS
4) Discuss the major complications of hEDS

Definition and Diagnosis

Ehlers-Danlos syndrome – Hypermobility Type (hEDS)

Criterion A -
Hypermobility on
Beighton score:
• > 6 for prepubertal children
• > 4 for males and females <50
• > 4 for females ages 50

Criterion B -
2 or more of:
• 1) Five or more of 12 connective tissue disorder signs
• 2) Positive family history
• 3) One of:
  • Persistent pain in 2 limbs or more for >3 mo
  • Chronic widespread pain for >3 months
  • Recurrent joint dislocations in the absence of trauma

Criterion C -
All
• Absence of unusual skin fragility
• Absence of other heritable connective tissue disorders
• Exclusion of other disorders

Joint Hypermobility
Beighton Score

> Hypermobility varies by:
  - Age: Young and old are more mobile than adults
  - Sex: Females > males
  - Ethnicity
  - Occupation/sports activities

5-Point Questionnaire for JHM

1) Could you ever bend your thumb to touch your forearm?
2) Could you ever bend your thumb to touch your forearm?
3) As a child did you amuse your friends by contorting your body into strange shapes or could you do the splits?
4) As a child or teenager did your shoulder or kneecap dislocate on more than one occasion?
5) As a child or teenager did you consider yourself double-jointed?

Agreement: affirmative answer for two or more questions.
Source: modified for Hakim and Grahame [2003]

Connective Tissue Disorder Signs

- Mild skin hyperextensibility
- Unexplained stria
- Bilateral pigmented papules of the heel
- Atrophic scarring in at least two sites not that of classic EDS
- Arachnodactyly (wrist or thumb sign)
- Arm span to height ratio ≤ 0.85
Connective Tissue Disorder Signs

- Unusually soft or velvety skin
- Recurrent or multiple hemias
- Pelvic floor, rectal or uterine prolapse without obesity or pregnancy
- Dental crowding with high or narrow palate
- Mitral valve prolapse
- Aortic root dilatation Z score >2

Differential Diagnosis -

**Marfan syndrome and related disorders**
- Aortic root dilatation, skin hyperextensibility, kyphoscoliosis, scoliosis, mitral valve prolapse, easy bruising, recurrent abdominal hernias, arachnodactyly
- Skeletal: slender, tall stature, long limbs, high palate, arachnodactyly
- Cardiovascular: aortic root dilatation
- Eye: ectopia lentis

**Loeys Dietz syndrome**
- Cardiovascular: aortic root dilatation, coarctation of the aorta, arterial tortuosity, hypertension
- Skeletal: craniofacial dysmorphisms, arterial tortuosity, hypertelorism
- Skin: redundant skin, cutis laxa
- Other: Marfanoid habitus, microcornea

**Arterial Tortuosity Syndrome**
- Cardiovascular: arterial tortuosity, aortic root dilatation, aortic regurgitation
- Skeletal: kyphoscoliosis, scoliosis
- Skin: redundant skin, cutis laxa
- Other: Marfanoid habitus, microcornea

Differential Diagnosis - Other

Other connective tissue disorders
- Mucocutaneous fragility, MAC, chronic myeloid leukemia
- Marfanoid habitus, ectopia lentis

Musculoskeletal disorder
- Musculoskeletal pain, atrophic scarring
- Cardiac failure

Rheumatological conditions
- Rheumatoid arthritis
- Ankylosing spondylitis
- Osteoarthritis

Neurological disorders
- Multiple sclerosis
- Amyotrophic lateral sclerosis
- Hereditary and acquired polyneuropathies

Consider EDS (any form) when:

- Late walking with joint hypermobility
- Abnormal bruising and bleeding
- Tissue fragility, atrophic scarring or skin hyperextensibility
- Symptomatic joint hypermobility with or without dislocations
- Unexplained vessel rupture or dissection
- Internal organ rupture

Natural History and Complications
JHS/EDS-HT
Multi-systemic disease
Wide inter- and intra-familial variability

Manifestations
- Mucocutaneous
- Skeletal
- Cardiovascular
- Gastrointestinal
- Gynecological
- Neurological
- Psychiatric

Natural history of JHS/EDS-HT is characterized by a progressive worsening:
- Usually mild in infancy
- More severe in the adulthood

Musculoskeletal
- Recurrent generalized dislocations
- Widespread chronic pain
- Scoliosis
- Soft-tissue lesions (i.e., bursitis, tenosynovitis)
- Pes planus
- Hypotonia
- Myofascial pain

Mucocutaneous
- Mildly hyperflexible skin
- Soft, silky, velvety texture
- Easy bruising
- Mast Cell Activation

Cardiovascular
- Orthostatic intolerance
- Mitral valve prolapse
- Valvular regurgitation
- Absent to mild aortic root dilation

Gastrointestinal
- GE Reflux
- Unexplained abdominal pain
- Constipation/Diarrhea/IBS
- Delayed Gastric emptying
Urogynecologic

- Urinary Stress incontinence
- Pelvic prolapse
- Dyspareunia

Neuropsychiatric

- Headaches
- Chronic Fatigue Syndrome
- Autonomic instability
- Recurrent parasthesias
- Depression
- OCD/Anxiety (RR >23)
- Sleep disturbances

Treatment of Manifestations

DO

- Regular, aerobic fitness
- Fitness support with strengthening, gentle stretching and proprioception exercises
- Postural and ergonomic hygiene especially during sleep, at school and workplace
- Weight control (BMI < 25)
- Daily relaxation activities
- Lubrication during sexual intercourse (women)
- Increased liquid intake (2-2.5L/day)
- High salt intake (except for arterial hypertension)
- Early treatment of malocclusion

DON'T

- High impact sports/activities
- Low environmental temperatures
- Prolonged sitting positions and prolonged recumbency
- Sudden head-up postural changes
- Excessive weight lifting/carrying
- Large meals (especially of refined carbohydrates)
- Hard foods, excessive jaw movements (ice, gums, etc.)
- Bladder irritant foods (e.g. coffee and citrus products) intake
- Nicotine and alcohol intake

Common Complications

- Orthostatic Intolerance
- Headaches
- Chiari Malformation
- Scoliosis/Kyphosis
- Abdominal Pain
- Pelvic Pain
Common Referrals

- Pain Management
- Physiatry
- Rheumatology
- Physical Therapy
- Sports Medicine
- Orthopedics
- Cardiology
- Neurosurgery
- Urology/Gynecology
- Gastroenterology

Joint Pain/Instability

- Strengthen surrounding muscle groups
- Elastic bracing as necessary
- NSAIDS, ice as necessary

Summary

- EDS hypermobile type is common
- Most other connective tissue disorders are less common
- Healthy lifestyle is the foundation to treatment
- This disorder is poorly understood but an active area of research

Skin

- Hyperextensibility

Molluscoid Pseudotumor Atrophic Scarring

https://www.hindawi.com/journals/isrn/2012/751768/fig1/

http://www.ehlersdanlosnetwork.org/vascular.html