Ehlers Danlos syndrom (EDS), både sällsynt och vanligt

Klassifikation (2017)

- 13 olika EDS subgrupper, 12 är sällsynta
- En liten bokstav används för att klargöra vilken subgrupp det handlar om (hEDS, cEDS, vEDS etc.)
- hEDS inte lika sällsynt, men utan genetisk markör
- HSD ingen ”light” variant till hEDS

När misstänks EDS/HSD?

- Hypermobilitet och ledinstabilitet
- Blödningsbenägenhet
- Hud/organ-skörhet
- Hud övertöjbarhet
- Ärftlighet
- Smärta

Time to diagnosis

The average time to diagnosis is more than 10 years

- The complex and often diffuse picture presented to sufferers and medical staff often leads to misdiagnosis and a long medical odyssey for EDS patients [31]. While pain is usually one of the first symptoms, the average time to diagnosis of the underlying disease is more than 10 years [32].

Diagnos HSD/hEDS

The International Consortium on Ehlers-Danlos Syndromes & Related Disorders

Diagnostic Criteria for Hypermobile Ehlers-Danlos Syndrome (hEDS)

This diagnostic checklist is for doctors across all disciplines to be able to diagnose EDS

Distributed by The Ehlers Danlos Society.

Patient name: _____ DOB: _____ DOV: _____ Evaluator: _____

The clinical diagnosis of hypermobile EDS needs the simultaneous presence of all criteria, 1 *and* 2 *and* 3.

CRITERION 1 – Generalized Joint Hypermobility

One of the following selected:

- ☐ ≥6 pre-pubertal children and adolescents
- ☐ ≥5 pubertal men and woman to age 50
- ☐ ≥4 men and women over the age of 50

Beighton Score: ____/9

If Beighton Score is one point below age- and sex-specific cut off, two or more of the following must also be selected to meet criterion:

- ☐ Can you now (or could you ever) place your hands flat on the floor without bending your knees?
- ☐ Can you now (or could you ever) bend your thumb to touch your forearm?
- ☐ As a child, did you amuse your friends by contorting your body into strange shapes or could you do the splits?
- ☐ As a child or teenager, did your shoulder or kneecap dislocate on more than one occasion?
- ☐ Do you consider yourself "double jointed"?

CRITERION 2 – Two or more of the following features (A, B, or C) must be present

Feature A (five must be present)

- ☐ Unusually soft or velvety skin
- ☐ Mild skin hyperextensibility
- ☐ Unexplained striae distensae or rubae at the back, groins, thighs, breasts and/or abdomen in adolescents, men or pre-pubertal women without a history of significant gain or loss of body fat or weight
- ☐ Bilateral piezogenic papules of the heel
- ☐ Recurrent or multiple abdominal hernia(s)
- ☐ Atrophic scarring involving at least two sites and without the formation of truly papyraceous and/or hemosideric scars as seen in classical EDS
- ☐ Pelvic floor, rectal, and/or uterine prolapse in children, men or nulliparous women without a history of morbid obesity or other known predisposing medical condition
- ☐ Dental crowding and high or narrow palate
- ☐ Arachnodactyly, as defined in one or more of the following:
  (i) positive wrist sign (Walker sign) on both sides, (ii) positive thumb sign (Steinberg sign) on both sides
- ☐ Arm span-to-height ratio ≥1.05
- ☐ Mitral valve prolapse (MVP) mild or greater based on strict echocardiographic criteria
- ☐ Aortic root dilatation with Z-score >+2

Feature A total: ____/12

Feature B

- ☐ Positive family history; one or more first-degree relatives independently meeting the current criteria for hEDS

Feature C (must have at least one)

- ☐ Musculoskeletal pain in two or more limbs, recurring daily for at least 3 months
- ☐ Chronic, widespread pain for ≥ 3 months
- ☐ Recurrent joint dislocations or frank joint instability, in the absence of trauma

CRITERION 3 – All of the following prerequisites MUST be met

1. Absence of unusual skin fragility, which should prompt consideration of other types of EDS
2. Exclusion of other heritable and acquired connective tissue disorders, including autoimmune rheumatologic conditions. In patients with an acquired CTD (e.g. Lupus, Rheumatoid Arthritis, etc.), additional diagnosis of hEDS requires meeting both Features A and B of Criterion 2. Feature C of Criterion 2 (chronic pain and/or instability) cannot be counted toward a diagnosis of hEDS in this situation.
3. Exclusion of alternative diagnoses that may also include joint hypermobility by means of hypotonia and/or connective tissue laxity. Alternative diagnoses and diagnostic categories include, but are not limited to, neuromuscular disorders (e.g. Bethlem myopathy), other hereditary disorders of the connective tissue (e.g. other types of EDS, Loeys-Dietz syndrome, Marfan syndrome), and skeletal dysplasias (e.g. osteogenesis imperfecta). Exclusion of these considerations may be based upon history, physical examination, and/or molecular genetic testing, as indicated.

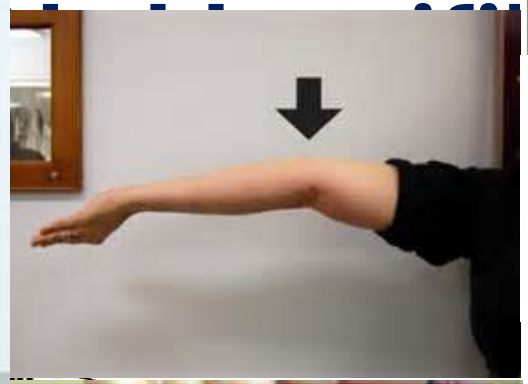
Diagnosis: _____

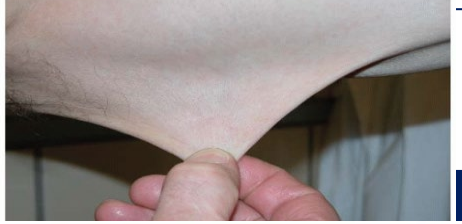
v9

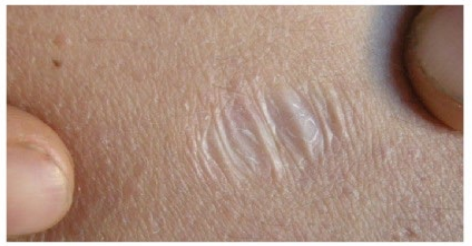
Kriteria 1

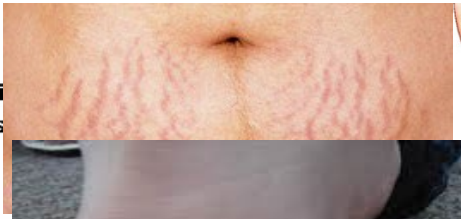
Kriteria 2 A

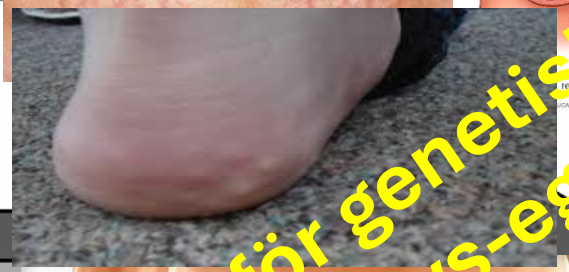
hEDS

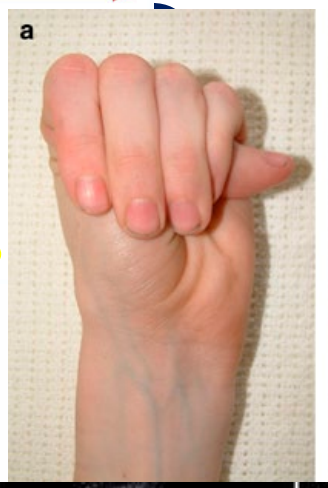


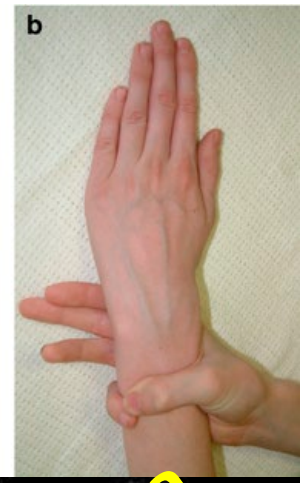


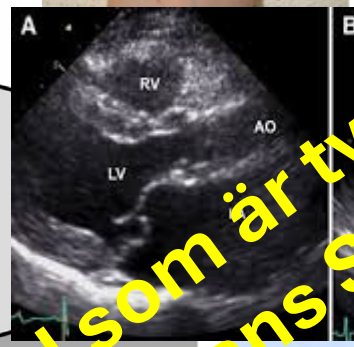


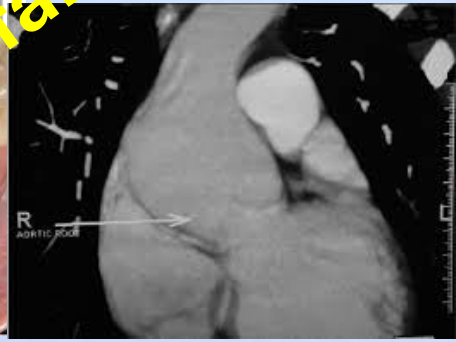












Beighton poäng

Uttryck för genetiskt avvikande bindväv-egenskaper

Fynd som är typiska vid Marfans Syndrom

EDS/HSD Kriteria 3

Arterial turtuosity syndrom
Loeys Dietz syndrom
Lateral meningocele syndrom
Bethlems myopati

Koagulationsrubbningar

Blödning

Fibromyalgi

Marfanoid utseende

Ostepeni

Ledsmärta

Marfan syndrom

Osteogenesis Imperfekta

Reumatiska sjukdomar

Diagnos HSD/hEDS

HSD

Ja

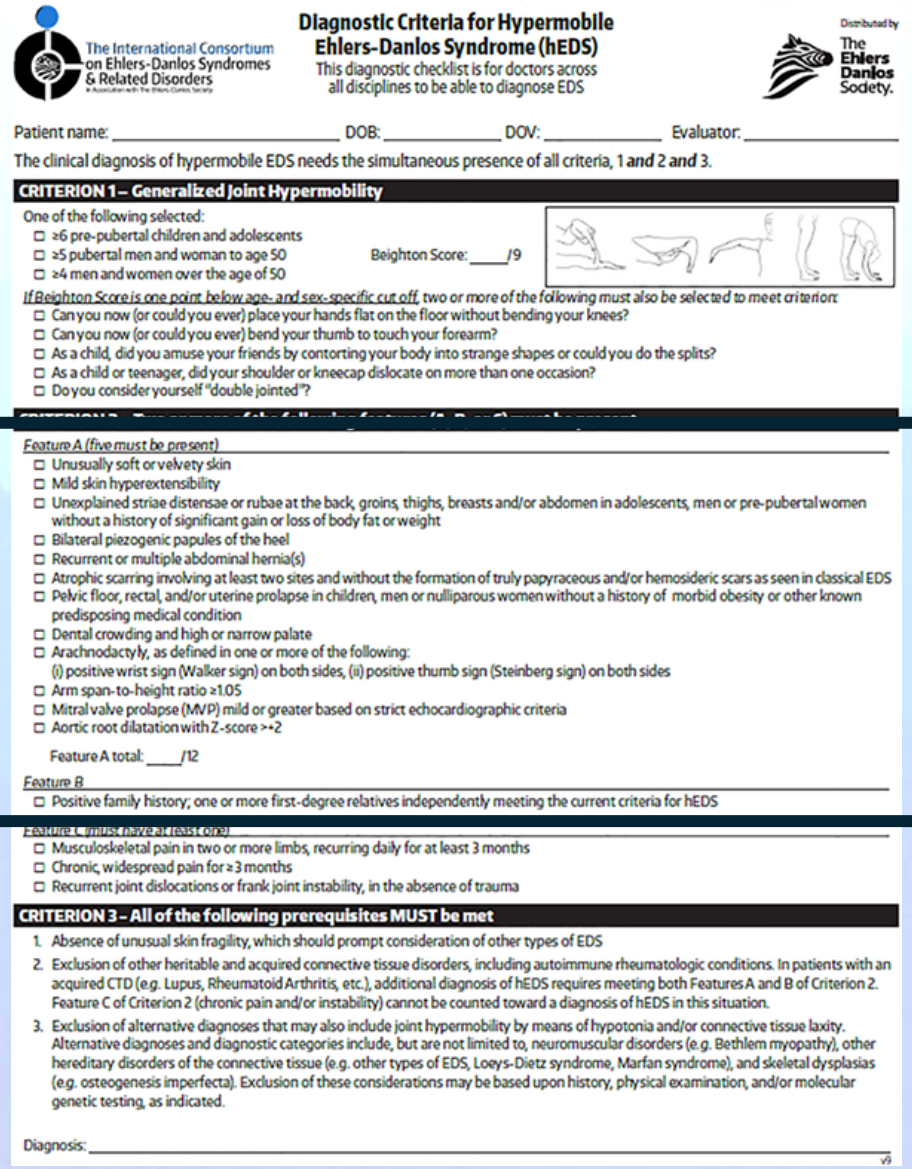
Under 5 poäng

nej

Ja

Ja

Ja



The International Consortium on Ehlers-Danlos Syndromes & Related Disorders

Diagnostic Criteria for Hypermobile Ehlers-Danlos Syndrome (hEDS)

This diagnostic checklist is for doctors across all disciplines to be able to diagnose EDS

Distributed by The Ehlers Danlos Society.

Patient name: _____ DOB: _____ DOV: _____ Evaluator: _____

The clinical diagnosis of hypermobile EDS needs the simultaneous presence of all criteria, 1 *and* 2 *and* 3.

CRITERION 1 – Generalized Joint Hypermobility

One of the following selected:

- ☐ ≥6 pre-pubertal children and adolescents
- ☐ ≥5 pubertal men and woman to age 50
- ☐ ≥4 men and women over the age of 50

Beighton Score: ____/9

If Beighton Score is one point below age- and sex-specific cut off, two or more of the following must also be selected to meet criterion:

- ☐ Can you now (or could you ever) place your hands flat on the floor without bending your knees?
- ☐ Can you now (or could you ever) bend your thumb to touch your forearm?
- ☐ As a child, did you amuse your friends by contorting your body into strange shapes or could you do the splits?
- ☐ As a child or teenager, did your shoulder or kneecap dislocate on more than one occasion?
- ☐ Do you consider yourself "double jointed"?

Feature A (five must be present)

- ☐ Unusually soft or velvety skin
- ☐ Mild skin hyperextensibility
- ☐ Unexplained striae distensae or rubae at the back, groins, thighs, breasts and/or abdomen in adolescents, men or pre-pubertal women without a history of significant gain or loss of body fat or weight
- ☐ Bilateral piezogenic papules of the heel
- ☐ Recurrent or multiple abdominal hernia(s)
- ☐ Atrophic scarring involving at least two sites and without the formation of truly papyraceous and/or hemosideric scars as seen in classical EDS
- ☐ Pelvic floor, rectal, and/or uterine prolapse in children, men or nulliparous women without a history of morbid obesity or other known predisposing medical condition
- ☐ Dental crowding and high or narrow palate
- ☐ Arachnodactyly, as defined in one or more of the following: (i) positive wrist sign (Walker sign) on both sides, (ii) positive thumb sign (Steinberg sign) on both sides
- ☐ Arm span-to-height ratio ≥1.05
- ☐ Mitral valve prolapse (MVP) mild or greater based on strict echocardiographic criteria
- ☐ Aortic root dilatation with Z-score >+2

Feature A total: ____/12

Feature B

- ☐ Positive family history; one or more first-degree relatives independently meeting the current criteria for hEDS

Feature C (must have at least one)

- ☐ Musculoskeletal pain in two or more limbs, recurring daily for at least 3 months
- ☐ Chronic, widespread pain for ≥ 3 months
- ☐ Recurrent joint dislocations or frank joint instability, in the absence of trauma

CRITERION 3 – All of the following prerequisites MUST be met

1. Absence of unusual skin fragility, which should prompt consideration of other types of EDS
2. Exclusion of other heritable and acquired connective tissue disorders, including autoimmune rheumatologic conditions. In patients with an acquired CTD (e.g. Lupus, Rheumatoid Arthritis, etc.), additional diagnosis of hEDS requires meeting both Features A and B of Criterion 2. Feature C of Criterion 2 (chronic pain and/or instability) cannot be counted toward a diagnosis of hEDS in this situation.
3. Exclusion of alternative diagnoses that may also include joint hypermobility by means of hypotonia and/or connective tissue laxity. Alternative diagnoses and diagnostic categories include, but are not limited to, neuromuscular disorders (e.g. Bethlem myopathy), other hereditary disorders of the connective tissue (e.g. other types of EDS, Loeys-Dietz syndrome, Marfan syndrome), and skeletal dysplasias (e.g. osteogenesis imperfecta). Exclusion of these considerations may be based upon history, physical examination, and/or molecular genetic testing, as indicated.

Diagnosis: _____

v9

hEDS

Ja

Över 5 poäng

Ja

Ja

Ja

Smärta vid EDS/HSD

Nociceptiv	Nociplastisk	Upprepad	Neuropatisk
Ledsmärta	”FM”	Subluxation	Central
Muskelsmärta		Luxation	Perifer
Inflammation			• Polyneuropati
			• Nervskada
			• Nervinklämmning

EDS/HSD

EDS/HSD-specifika symtom

1. Hypermobilitet/ledinstabilitet
   - Smärta
   - ledsymtom
     - Låsningar, subluxation, luxation
2. Hudsymtom
   - Skörhet, läkningsproblem, breda ärr
3. Blödningsbenägenhet
   - Förlängd blödning, blåmärken

Komorbiditet

- Trötthet
- Sömnproblem
- IBS-liknande symtom
- MCAS
- Autonom dysfunktion
- PMS
- Dysparuni
- (Infektions känslighet?)

”Jag har blivit sämre i min HSD/EDS!”

Behandling och rehabilitering

Smärtbehandling vid EDS

- Framgångsrik behandling av långvarig smärta vid EDS ska vara multidisciplinär enligt så kallad ”bio-psyko-social” modellen
- Fysioterapi framförallt proprioceptiv träning ger långvarig symtomlindring *[Palmera et al., 2014]. [Grahame, 2009; Rozen, 2014]. [Engelbert et al., 2017].*

Behandling/rehabilitering

EDS/HSD

- Fysioterapi/arbetsterapi
  - Proprioceptiv träning
  - Enteroceptiv ”träning”
  - Hjälpmedel
- Läkemedel
  - Nociceptiv smärta
  - Neuropatisk smärta
- Kirurgi

Komorbiditet

- Egenvård
  - Vatten, stödstrumpor, salta extra på maten, röra på sig
- H1, H2, H3-receptorblockad
- Antiarytmiska läkemedel
- Dropp
- Syrgas
- Botulinum toxin

EDS/HSD smärt-orsaker och behandling

Smärta	Defenition	Debut	orsak	Behandling
Nociceptiv	Vävnad skada	Tidig ålder	Led/muskel-skada	Fysioterapi, arbetsterapi, läkemedel
Neuropatisk	Skada eller sjukdom i nervsystemet	Senare debut	Ändring i nervsignal-överföring	Läkemedel
Nedsatt proprioception	Nedsatt ledsinne	Tidig eller senare debut	Skada av proprioceptiva receptorerna	Fysioterapi, arbetsterapi

An official website of the United States government
View full-text article in PMC
. 2023 Mar 24;11(7):936. doi: 10.3390/healthcare11070936

Farmakologisk behandling av neuropatisk smärta (NeP), och nociceptiv smärta

1.NeP:

- TCA,
- AEP,
- SNRI,
- lokal applikation av Lidokain eller Capsaicin kan ge symtomlindring [Hamonet and Brock, 2015].
- Botulinum Toxin, klonidin, LDN

2.Nociceptiv smärta:

- Paracetamol,
- lokalinfiltration med Lidokain eller Botulinum Toxin [Hamonet et al., 2014; Hamonet and Brock, 2015].

Anpassad aktivitet


Tack