Ehlers-Danlos Syndrome (EDS)
For Healthcare Providers

This is a customized health care provider version of our website. Please visit the main website to find more comprehensive information for families and schools (www.gemssforschools.org).

Ehlers-Danlos (EDS) is a name given to a group of connective tissue disorders. This group includes a number of types, and was most recently reviewed by an International Consortium who released the 2017 Guidelines for diagnosis and treatment in March 2017. Using these guidelines, Hypermobility Type EDS (or hEDS) is generally considered the most common and the mildest type of EDS. It is also the least well-defined. The diagnosis of hEDS is based entirely on clinical evaluation and medical and family history, as there is no genetic testing available for confirmation. There is genetic testing available for the Classical and Vascular types that are much less common. Three of the types of Ehlers-Danlos syndrome are included here.

Physical characteristics and/or symptoms

Note: not all people with any of the types of EDS will have all of these features.

Hypermobile Ehlers-Danlos (hEDS) – the most common type

Diagnostic challenges
Joint hypermobility is more common in young children making the diagnosis more difficult in young children particularly in the absence of a family history. The 2017 International Consortium Guidelines for hEDS state that in order to make a diagnosis of hEDS, the patient needs to meet **ALL** of the following criteria:

CRITERIA 1: Generalized joint hypermobility (GJH)

1. Joint hypermobility, using the Beighton scale
   - ≥ 6 in a prepubertal child or adolescent
   - ≥ 5 in a pubertal men and females to the age of 50
   - ≥ 4 in individuals over the age of 50
2. If within one point of the above cutoff values, a historical 5 point questionnaire can be used

CRITERIA 2: Must include two of the following features:

1. Feature A: Findings of a generalized connective tissue disorder

- These include skin findings such as soft skin, hernias, and cardiac valvular findings

2. Feature B: Positive family history
3. Feature C: Musculoskeletal complications
    - These include joint dislocations, and musculoskeletal pain

CRITERIA 3: Prerequisites - all must be met

1. Absence of significant skin fragility
2. Other connective tissue disorders must be ruled out
3. Alternative reasons for joint hypermobility such as hypertonia or a feature of a syndrome must be ruled out

There are other findings that may support the diagnosis of hEDS, but are not sufficient to make the diagnosis. These can include:

- Functional bowel disorders
    - Junctional gastritis
    - Irritable bowel syndrome
- Postural orthostatic tachycardia (POTS) or dysautonomia
- **NOTE: Increased bone fragility and fractures are NOT part of hEDS in children and only occur in adults with osteopenia, usually related to decreased mobility and other causes**

Treatment of EDS-HT remains symptomatic.

- Physical therapy tailored to the individual
- Assisted devices and
- Pain management
- Low-impact weight-bearing exercise is imperative in order to improve joint stability and reduce the risk of osteopenia.

Classical Ehlers-Danlos (cEDS)

- Skin problems
    - Soft velvet like skin
    - Fragile skin that bruises or tears easily
    - Stretchy rubber band-like skin
    - Easy bruising, can be severe
    - Poor and slow wound healing
    - Small harmless bumps under skin
- Joint problems
    - Loose, unstable joints causes dislocation

    - Hyper-extensible joints
    - Joint pain
    - Temporomandibular joint (TMJ) laxity and pain
- Eye problems
    - Nearsightedness
- Postural orthostatic tachycardia (POTS) or dysautonomia

Vascular Ehlers-Danlos (vEDS) – much less common and not comprehensively addressed on the GEMSS website

Typical Features include:

- Characteristic facial appearance (seen in only some of the individuals)
    - Thin lips, small chin, thin nose, large eyes
- Skin problems
    - Thin, translucent
    - Easy bruising
    - Early onset varicose veins
    - Aged appearance, especially in hands
- Joint problems
    - Hypermobility of small joints
    - Chronic subluxations/dislocations
    - Congenital hip dysplasia
    - Clubfoot (in some)
- Vascular
    - Arterial rupture
    - Intestinal rupture
    - Uterine rupture during pregnancy
    - Abnormal connections between arteries and veins

Recommended Routine Surveillance

- Annual echocardiograms if cardiac findings are present
- Annual ophthalmologic exams
- Management of joint laxity and joint instability
- Pain management
- Skin fragility prevention as needed
- Evaluation for postural orthostatic tachycardia (POTS) or dysautonomia
- Psychosocial evaluation
- In vEDS, the routine surveillance will be dictated by the specialists involved

Emergency Protocols

There are no specific emergency protocols for hEDS. However, for a child with cEDS or vEDS, the school needs to be aware of the type of medical complications that can occur with injuries.

- Emergencies should be handled as with any child.

Specialists Who May Be Involved

The specialists that should be involved is based not only on the type of EDS, but also the severity of the individual case.

For all types:

- Cardiologist
  - Mitral valve prolapse
  - Autonomic dysfunction
  - Raynaud syndrome and acrocyanosis
- Dentist
  - TMJ syndrome
  - Dental crowding
- Dermatologist
  - Scar prevention
  - Molluscoid pseudotumors
- Gastroenterologist
  - IBS
  - Diarrhea and constipation
  - GERD
  - Junctional gastritis
- Geneticist / Genetic Counselor:
  - Diagnosis
  - Coordination of care
  - Genetic risk for family
  - Clinical trials
- Neurologist
  - Postural hypotension
  - Headache
  - Poor balance
  - Neuropathic pain
- Ophthalmology
  - Myopia
- Orthopedist

    - Joint laxity /instability
    - Osteoarthritis and osteoarthritis pain
    - Iliotibal band syndrome
    - Tendinitis and bursitis
    - Muscle or myofascial pain
- Pain therapist
- Physical therapist
- Psychologist
    - Psychological impairment and dysfunction
    - Emotional problems

Sample Forms

- Sample paragraph to be used for Letters of Medical Necessity or Letters to the school:

My patient_____ has been diagnosed with Classical Ehlers Danlos Syndrome. Classical EDS (cEDS) is characterized by skin hyperextensibility and fragility, abnormal wound healing, and joint hypermobility. Medical complications include management of joint hypermobility including frequent joint dislocations, hypotonia, fatigue, muscle cramps chronic pain, and easy bruising. Because of these, _____ needs the following accommodations.

My patient_____ has been diagnosed with Hypermobile Ehlers Danlos Syndrome. Hypermobile EDS (hEDS) is characterized by mild skin hyperextensibility, and joint hypermobility. Medical complications include management of joint hypermobility including frequent joint subluxations/ dislocations, joint pain, and autonomic dysfunction .Because of these, _____ needs the following accommodations.

Seven Important Aspects of School Life

"Ehlers-Danlos Syndrome at a Glance" will help you talk with parents and schools about:

- Medical / Dietary Needs
- Education Supports
- Behavior & Sensory Supports
- Physical Activity, Trips, Events
- School Absences & Fatigue
- Emergency Planning
- Transitions

Resources

GeneReviews - Hypermobile EDS
http://www.ncbi.nlm.nih.gov/books/NBK1279/
Gene Reviews: Ehlers-Danlos, Hypermobility Type

GeneRewiews – Classical EDS
http://www.ncbi.nlm.nih.gov/books/NBK1244/
Gene Reviews: Ehlers-Danlos, Classic Type

Ehlers-Danlos National Foundation
http://www.ednf.org
Hypermobile EDS Criteria Checklist
https://ehlers-danlos.com/wp-content/uploads/hEDS-Dx-Criteria-checklist-1.pdf