

Ehlers-Danlos Syndrome (EDS) at a Glance

The Ehlers Danlos Syndromes (EDS) is a group of hereditary connective tissue disorders that primarily affect the skin and joints. Connective tissue provides support to many parts of body like skin, muscles, and ligaments. People with EDS may have fragile skin or unstable joints.

- Marfan syndrome is another disorder of the connective tissue.

About 1 in every 20,000 babies is born with EDS. It can be inherited in an autosomal dominant, autosomal recessive, or X-linked recessive manner.



Meet Bayla on page 9

NOTE: The types of EDS were reviewed and new classifications and criteria were published in March of 2017. These pages reflect those changes.

There are 13 recognized types of EDS in the new classification. The new names include a descriptive name followed by Ehlers-Danlos syndrome:

- The most common type is Hypermobile EDS (hEDS), formerly known as Type III;
- Followed by Classical EDS (cEDS), formerly known as Type I and
- Vascular EDS (vEDS), formerly known as Type IV.

The types are distinct from each other not only in their genetic causes, but also in their symptoms. It is important to know which type has been diagnosed in the child. The type will hold true in families. This means that if one member of the family has Hypermobile EDS, the other members who have the condition will have that type, not another type. Within each type, the severity of the symptoms can vary between individuals even in the same family.

Things to Think About

1. Medical/Dietary Needs

What you need to know

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No special diet is required for EDS, although a well-balanced diet is important.

The list of *possible* medical problems in each type of EDS can be extensive. However, each individual usually has only some of these problems. Also, the severity of any one of these medical problems varies widely between individuals. Therefore, it is important to ask the parents about the medical issues in their child.

School age children with EDS may have annual doctor and specialist visits to monitor medical conditions.

Physical characteristics and/or symptoms:

Hypermobile EDS – the most common type:

- GENERALIZED JOINT HYPERMOBILITY IS AN ESSENTIAL FEATURE
 - Joint hypermobility is common in children without EDS
- Other joint problems
 - Recurrent dislocations/subluxations (altered position or partial dislocation)
 - Joint pain
 - Temporomandibular joint (TMJ) laxity and pain
- Skin problems
 - Soft velvet like skin
 - Stretch marks not related to weight gain
 - May have some easy bruising
 - NOT associated with fragile skin
- Other findings
 - Chronic pain – can be joint-related or can be a burning type of pain
 - Functional bowel disorders
 - Irritable bowel syndrome
 - Postural orthostatic tachycardia (POTS) or dysautonomia
 - This is the presence of excessive tachycardia (faster than normal heart rate) and other symptoms upon standing.
 - If you are interested in additional information on POTS/dysautonomia please see: <http://www.dinet.org/content/information-resources/pots/pots-symptoms-r96/>
 - High, narrow palate (roof of the mouth)
 - Dental crowding
 - NOT associated with increased fractures in children

Classical EDS

- Skin problems
 - Soft velvet like skin
 - Fragile skin that bruises or tears easily

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- 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 (from dislocation)
 - Temporomandibular joint (TMJ) laxity and pain
- Eye problems
 - Nearsightedness
- Postural orthostatic tachycardia (POTS) or dysautonomia
 - This is the presence of excessive tachycardia (faster than normal heart rate) and many other symptoms upon standing.
 - If you are interested in additional information about POTS/dysautonomia please see:
<http://www.dinet.org/content/information-resources/pots/pots-symptoms-r96/>.

Vascular EDS – much less common

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

What you can do

- A yearly check-up and studies as needed should occur in the child's Medical Home.
- Discuss pain management plan with parents
 - Use anti-inflammatories as appropriate
 - Allow storage of ice pack/gel packs at school
 - Avoid aspirin
- Individuals may complain of frequent stomach-aches due to reflux, delayed emptying,

irritable bowel, etc.

- A plan should be in place for dealing with these symptoms
- It is important to address injuries quickly. Children with dysautonomia may not report pain.
- Regular eye exams
- Be aware of any changes in behavior or mood that seem out of line with the situation and notify the parents.
- It is important to be aware of any academic changes. Contact parents when any differences are noticed.

2. Education Supports

What you need to know

It is important to have HIGH LEARNING EXPECTATIONS for children who have Ehlers-Danlos syndrome. Encourage use of the core educational curriculum and modify it in order to meet the individual needs of the child.

There is no evidence that EDS in itself causes learning complications. However, premature birth is a complication associated with EDS and this may cause learning differences.

Individuals may have speech, visual, hearing problems. There also may be motor difficulties due to joint hypermobility.

What you can do

- Formal IQ and performance testing may be useful.
- If there are verbal performance discrepancies, make sure it is not because of motor difficulties
 - Motor difficulties due to hand hypermobility can affect written test scores.
 - May require alternative ways to assess performance or allow extra time
- Pair student with a note taker
- Use pencil grips
- Some individuals with Hypermobility EDS also find finger ring splints to be helpful.
- Use Computers
- Provide two sets of textbooks: one for home and one for school
- Allow the use of an elevator and/or extra time to go to and from classrooms
- Provide a locker at eye level with a digital lock
- Offer priority seating
- Provide a cushion for sitting on the floor and for a chair seat
- Music can be helpful
 - Select instruments to minimize stress to joints

- If POTS (postural orthostatic tachycardia) is present
 - Provide extra fluids prior to rising from a rest period
 - Allow frequent breaks as sitting for a long time in one position is difficult

3. Behavioral and Sensory Support

Individuals with EDS may have:

- Psychological and psychosocial difficulties
 - Fatigue and pain may make psychological problems worse
 - Psychological distress may increase pain
 - Depression, anxiety, affective disorder, low self-confidence, negative thinking, hopelessness and desperation may occur
 - Depression is often associated with chronic pain, disabilities
 - Individuals may feel misunderstood, disbelieved, or alone
 - Children may be self-conscious of physical differences
- Pain
 - Pain can be a major part of EDS
 - Fear of pain and joint instability may lead to avoidance behaviors, and make dysfunction and disability worse

What you can do

- Counseling and support for pain may help
- Meditation and yoga could be helpful
- Antidepressants may be helpful
- Need help with adaptation and acceptance of issues and potential limitations.

4. Physical Activity, Trips, Events

What you need to know

EDS is different for each person. An individual with EDS may require no accommodations or many. Individuals may require a 504 plan.

An alternative health related credit should be provided if PE accommodations are not possible. Children with dysautonomia may have difficulty in extreme temperatures and may require air conditioning on very hot days and may need to avoid very cold temperatures.

With Hypermobile EDS:

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- Learning to protect the joints will help prevent further injury and keep individuals active
- Individuals should participate in activities that do not cause joint pain and finding less painful ways to move and perform certain tasks
- Avoiding activities that overextend or lock joints
- Frequent overextension of joints can cause traumatic arthritis
- Joint stability can be improved through exercise programs to strengthen muscles
- Low resistance exercise may increase muscle tone
- Important to increase both core and extremity strength
- Position sense may be off in some of these individuals
- If finger and hand strain occurs, may need writing adaptations
- Non weight bearing exercise promotes strength and coordination

What you can do

Non weight bearing exercise

- Swimming/water exercises
- Walking
- Biking
- Low impact aerobics
- Core toning exercises

Physical Therapy

- Myofascial release provides short term relief of pain
- Heat, cold, massage, ultrasound, electrical stimulation, acupuncture, biofeedback, and relaxation also may help
- Low resistance muscle toning exercise may improve joint stability
- Exercises that promote improved proprioception (position sense) or balance are important

Assistive devices

- Braces to improve joint stability
- Wheelchairs/scooter may help relieve stress
- Suitable mattress to improve sleep
- May wear soccer pads or ski stocking to protect skin from bruising during activities

Pain medication

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- Mild to moderate:
 - As needed medication may be sufficient
- More significant pain
 - May require higher doses and multiple medications
- Prevention and control of pain is important

Occupational Therapy

- Teach joint protection strategies
- Adjust chair/desk
- Ring splints to stabilize finger joints
- Wrist or wrist/thumb braces for small joint instability
- Neck collar
 - May help with neck pain and headaches
- Wheelchair/scooter
- May require accommodations in school
 - Use of elevator
 - Extra time in halls

Avoid the following:

- Joint hyperextension
- Resistance exercises which can make joint instability and pain worse
 - Resistance bands may not be effective
- Isometric exercises, which can be problematic with too much resistance
- High impact activity increases risk for
 - Subluxation/dislocation
 - Chronic pain
 - Osteoarthritis
- Chiropractic adjustment with caution
- Crutches and canes may put increased stress on upper extremities
- Certain activities
 - Weightlifting
 - Running
 - Contact/ fighting sports
 - Football
- May not be able to lift books/heavy backpacks

Note: With Classical EDS there will need to be a much higher vigilance for injury because of the skin fragility.

5. School Absences and Fatigue

What you need to know

- Chronic absences may be an issue
- Joint pain and instability may cause fatigue
- Contact parents if changes are noticed

What you can do

- Provide assignments and notes
- Assistive devices may help
- Frequent breaks are important

6. Emergency Planning

What you need to know

- Develop an emergency plan if necessary, depending on the needs of individual children
- Specific emergency plans may be required in case of cuts, subluxation, dislocations, or internal problems especially with EDS – vascular type
 - See www.ednf.org for urgent information on vascular complications

7. Resources

GeneReviews – Vascular Type

<https://www.ncbi.nlm.nih.gov/books/NBK1494/>

Gene Reviews: Ehlers-Danlos, Vascular Type

GeneReviews – Hypermobility EDS

<http://www.ncbi.nlm.nih.gov/books/NBK1279/>

Gene Reviews: Ehlers-Danlos, Hypermobility Type

GeneReviews – Classical EDS

<http://www.ncbi.nlm.nih.gov/books/NBK1244/>

Gene Reviews: Ehlers-Danlos, Classic Type

2017 International Classification of the Ehlers-Danlos Syndrome

<http://onlinelibrary.wiley.com/doi/10.1002/ajmg.c.31552/epdf>

<https://www.ehlers-danlos.com/2017-eds-international-classification/>

Medline Plus

<https://medlineplus.gov/genetics/condition/ehlers-danlos-syndrome/>

Consumer-friendly information about human genetics from the U.S. National Library of Medicine

Ehlers-Danlos Society

www.ehlers-danlos.com

Our volunteers and our families are the lifeblood of our mission.

http://ednf.org/sites/default/files/EDNF_ParentsGuideForEducators.pdf

Educator's Guide: Meeting the Needs of the Ehlers Danlos Child / Parent's Guide: Helping Your Child Succeed at School

School Issues with Ehlers-Danlos syndrome

<http://www.ehlersdanlosnetwork.org/School-Issues.html>

Five Frequently Asked Questions about EDS and School by Peggy Rocha Snuggs

8. Meet a Child with Ehlers-Danlos Syndrome

Being Bayla

GEMSS would like to thank Bayla and her mother for their generosity in sharing this story with us. You have made the site come to life with the addition of your thoughts and feelings. Thank you so much!

The pieces of the puzzle started to come together for Bayla and her family after little Bayla tripped and fell on a family camping trip. This resulted in an extensive injury which probably would not have been serious in most children. A plastic surgeon was needed to repair the injury, and he suggested a follow up with her physician because of the extensiveness of the injury and the great elasticity of her skin. The plastic surgeon remembered learning about Ehlers-Danlos Syndrome in medical school but never had



seen anybody with it. He felt her symptoms aligned with the diagnosis of Ehlers-Danlos Syndrome. Further investigation with a genetics team confirmed the suspicion and a blood test showed that she had the ‘classic type’ of the syndrome (there are 5 other types). Suddenly, all the bruises that appeared from small events, the significant scarring after cuts, and her overly frail skin made sense with this new diagnosis.

Bayla is a vibrant, fun-loving four-year-old who her mother describes as “outgoing, sassy, and full of life!” She will soon be starting kindergarten. Plans are underway to make sure the staff knows Bayla’s needs, her symptoms, and her abilities well before the first day of school. Bayla had been in preschool for a short while. However, her mother Kate felt home-school communication needs were so important and her own anxiety was high with this new diagnosis, so Bayla withdrew. Kate notes how important it was to hear *every* physical complaint Bayla made in a timely way so her medical needs could be addressed. This experience makes Kate a little nervous about kindergarten but she wants to work hard to make sure everyone is on the same page.

Bayla loves to play sports and, with this diagnosis, contact sports pose more of a risk for injury. Though Bayla has to choose sports like swimming that have less direct contact, her family encourages her to explore all that interests her. Bayla and her family take the precautions needed to keep her body free from injury without limiting the exploration of new activities. Bayla’s family wants to make sure she can experience a full and active life. Bayla’s diagnosis also gives her an edge in some areas! She is much more limber and flexible than the average person. Sprains, dislocations, bruising, and skin tears happen often with

Bayla. For most parents, this would be associated with a lot of anxiety, but Bayla's parents realize that this is their "new normal" for life with Bayla and do not panic with every injury. They have learned to be comfortable with managing whatever injury may arise and any care she may need.

There is anxiety that goes along with sending Bayla off to play in the care of someone else but Kate makes sure everyone knows about Bayla's condition. She is careful not limit Bayla's ability to "spread her wings and fly." Bayla has two siblings who know that she must be treated a little more gingerly. Her parents have helped her older sister learn that Bayla is more sensitive and, consequently, her sister has developed a good sense of what might be dangerous.

Kate is a nurse administrator and has some solid advice and strategies for both parents and teachers.

For parents:

- Trust your instincts as a parent! Kate felt as if something was not right 2 years prior to the diagnosis. She had questioned the significant bruising, thin skin, and prolapsed rectum at her wellness visits but the work ups did not happen until sometime later.
- Let your child find their own boundaries. Know that as they age, they will become more knowledgeable about their bodies and aware of potential challenges and restrictions.
- Make connections with others who have EDS. The best resource Kate has found are those people who are older (even adults) and have lived with this syndrome. They have offered great tips for how to preserve and protect Bayla's body and keep it free from injury.
- Spread awareness, to doctors, friends, and family. Bayla's parents have found that there are many people, including physicians, that are not aware of this syndrome.

For teachers:

- Keep the lines of communication open with the child and parents so that everyone is aware of what is happening. Watch for cues and listen to the child's complaints with care.
- Report even minor complaints to the parents as they are monitoring this carefully.
- Children with EDS have faulty collagen in their body. They bruise more than other children and are prone to greater injury than other children. A simple grab of the

arm as a child runs away can result in a severe bruise. Be aware of this and make sure the children playing with a child with EDS know this as well. Classmates need to know that play has to be gentle.

- Working with a child with EDS should be a collaborative approach in the school setting. The teachers on the playground, the gym teacher, the school nurse, and the child's classmates all need to be aware of the child's health condition.