

AGE 12+ RECOMMENDED YEARLY SCREENING

Connective Tissue Disorders & Ehlers-Danlos Syndrome

Collagen is the most abundant protein in the human body. It provides structural strength in human tissues, including heart and blood vessels, eyes and skin, cartilage and bone. When muscles, ligaments, tendons and even large organs are built with structurally defective collagen, there can be systemic weakness and instability evident throughout. Connective tissue disorders such as Ehlers-Danlos syndrome cause multiple manifestations that affect many functions of the human body.

Why Is Screening Important?

Children with a connective tissue disorder will usually display warning signs before any serious problems begin to arise. Recognizing these warning signs early provides the best opportunity to positively influence the course of patients' lives. Families with Ehlers-Danlos syndrome should be educated to be aware of the potential risks of contact sports, and how to preserve already unstable joints and general health.

What Can I Do?

The most common form of EDS is hypermobility, which is the form for which genetic testing is unavailable (the genes involved have not been completely identified yet). So it is particularly important that primary care physicians screen for EDS. Refer the family to a geneticist, but:

You are in the best position to enable an EDS child's full and happy life.

The clues and complications listed here can help guide you and the families you serve in deciding whether a diagnosis of EDS may be worth pursuing further, and help those who have been diagnosed to stay as healthy as possible.

Visit EDNF.org to find further information for both families and medical professionals.

CARDIOVASCULAR

- Possibility of aortic root dilatation, mitral valve prolapse, other valvular abnormalities, enlarged right coronary artery.
- Postural orthostatic tachycardia, leading to chronic fatigue, is especially found in young persons with EDS.
- Lipid abnormalities, onset at a young age.

GASTROENTEROLOGY

- Irritable bowel syndrome with constipation and/or diarrhea, reflux, food allergies, gastroparesis.

RHEUMATOLOGY & ORTHOPEDIC

- Joint hypermobility can be assessed using the Beighton scale; however, joint hypermobility also depends on age, gender, family and ethnic background.
- Excessive flattening of feet when weight bearing, pronated or everted feet, plantar fasciitis, bunions.
- Joint dislocations & subluxations apparently unrelated to specific injury.
- Chronic unexplained joint pain, commonly out of proportion to physical and radiological findings.
- Scoliosis, kyphosis and leg length discrepancy, knee/hip alignment issues.
- Premature onset of degenerative disc disease and herniated discs in the spine.

SOCIAL & DEVELOPMENTAL

- Depression and withdrawal from social activities due to chronic pain.
- Physical awkwardness and clumsiness.

DERMATOLOGY & SPORTS MEDICINE

- Easy bruising, enlarged scars, stretch marks, poor wound healing.
- Frequent injuries.
- Joints may not be stabilized by adequate muscular control and appropriate physical therapy.
- Long-term damage resulting from hypermobile joint "party tricks", rotational stress, contact sports.

MAXILLOFACIAL/DENTAL

- High palate and teeth crowding (prior to orthodontic corrections).
- TMJ pain.
- Early onset gingival recession and gum problems.
- Cavities, dental discoloration and dental pits.

NEUROLOGY

- Chiari I malformation, cranio-cervical instability, cervical disc disease.
- Syringomyelia.
- Tethered Cord Syndrome.
- Migraine headaches.