EHLERS-DANLOS SYNDROME

Collagen is the most abundant protein in the human body.

It provides the structural strength in most human tissue, including the heart and blood vessels, eyes and skin, cartilage and bone.

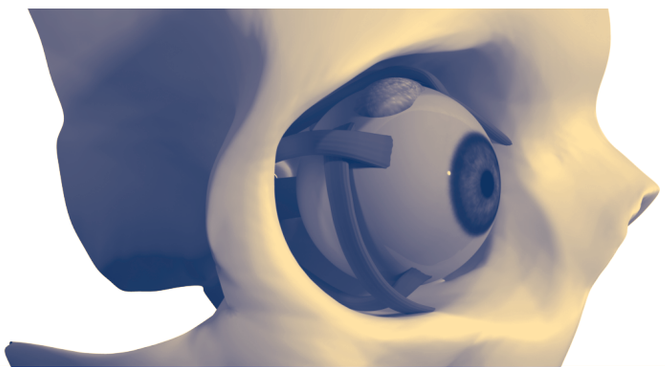
What happens when this basic building block is flawed?

When muscles, ligaments, tendons and even large organs are built with structurally defective collagen there is systemic weakness and instability evident throughout the body.

There is Ehlers-Danlos Syndrome.

Excessively mobile joints, chronic pain and skin softness characterize Ehlers-Danlos syndrome (EDS). At least six types of EDS have been identified; clinical manifestations vary according to type and may also include high myopia, photophobia, poor wound healing with atrophic scars, easy bruising, and generalized connective tissue fragility. Each type is thought to involve a unique defect in connective tissue, although not all of the genes responsible for causing EDS have been found. Within each family the type of EDS runs true, but individual family members may vary in clinical severity and manifestations.

As a group of genetic disorders of connective tissue, the estimated prevalence of EDS is 1 in 5,000. It is known to affect men and women of all racial and ethnic backgrounds.



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OPHTHALMOLOGY & EDS

The most recognizable ophthalmological consideration in EDS is the very rare Kyphoscoliosis type, addressed at right. But considering the sclera and cornea are mostly collagen, patients with EDS or any collagen-related disorder should be carefully watched for ocular disease. Ophthalmological abnormalities in EDS include but are not limited to myopia, retinal detachment and glaucoma. Some of these can be vision-threatening.

An optometrist or ophthalmologist should establish a comprehensive baseline for a person with EDS, including a complete eye exam and history. Annual follow-ups should then reveal any changes.

Baseline Eye Exam Recommendations for EDS

- Complete slit lamp exam with TBUT;
- Dilated fundus examination with photography;
- Ocular topography;
- Scanning Laser Ophthalmoscopy: Retinal Thickness Analyzer, Stratus OCT, *etc.*;
- Orbscan and/or pachymetry to check corneal thickness;
- Pupil testing, aperture measurements;
- EDS patients are not good candidates for LASIK.

Any disturbance in vision or eye health should be evaluated and explained, particularly in the presence of collagen disorders. Patients with retinal change should have follow-up visits every six months, and patients who report floaters should be evaluated every three months. Anyone experiencing flashes of light should immediately get in touch with their eye-care physician.

Indicators and symptoms include:

Eyelid laxity, blue sclera, photophobia, thin cornea, high myopia, posterior staphyloma, strabismus, angioid streaks, dry eyes.

Potential serious concerns:

Lens subluxation, keratoconus, cataracts, glaucoma, macular degeneration.

Emergency concerns:

Carotid-cavernous sinus fistula (especially in Vascular EDS patients) , retinal detachment.

KYPHOSCOLIOSIS TYPE INFORMATION

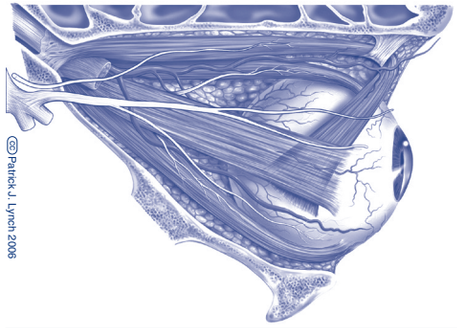
The Kyphoscoliosis type of EDS (previously Type VI) is specifically caused by a deficiency of lysyl hydroxylase, an enzyme essential for the assembly of collagen fibrils; inheritance is autosomal recessive. Effects may include a progressive loss of eye pigment tissue; glaucoma, promoted by an improper drainage of eye fluid leading to higher intraocular pressure; and scleral fragility, including ocular rupture.

Major Diagnostic Criteria

- Joint laxity
- Severe hypotonia at birth
- Scoliosis, progressive
- Scleral fragility or rupture of globe

Minor Diagnostic Criteria

- Tissue fragility
- Easy bruising
- Arterial rupture
- Marfanoid features
- Microcornea
- Osteopenia
- Positive family history (affected sibling)



FOR THE EDS PATIENT: EYE COMPLAINTS & SYMPTOMS

These problems are not necessarily related to EDS, but they require a more intense evaluation in someone with EDS.

- Blurred vision that comes and goes; seeing double;
- Complete/almost complete vision loss in one eye lasting for minutes; migraine auras;
- Dry eyes, light sensitivity, tunnel vision, floaters.

SEEK EMERGENCY HELP IF ANY OF THESE OCCUR:

- Double vision that starts suddenly, especially if vertical;
- Flashes of light (with or without floaters);
- Pain, redness or discharge;
- Curtain coming up over vision;
- Frontal headache in which patient "hears pulse in the temple";
- Sudden change in vision.

URGENT INFORMATION on Vascular EDS

CAROTID-CAVERNOUS FISTULA IS A LIFE-THREATENING EMERGENCY.

Redness, pain and prominence of one or both eyes and the sound of pulsations in the head can indicate a carotid-cavernous fistula. High pressure blood from the internal carotid artery can pass directly into veins behind the eye, potentially leaking out of the confines of the blood vessels to become life-threatening. Seek immediate hospital-based medical attention, and inform emergency medical staff of Vascular EDS and the risk of carotid-cavernous fistula.

ARTERIAL RUPTURE IS THE MOST COMMON CAUSE OF SUDDEN DEATH.

- ***Arterial or intestinal rupture commonly presents as acute abdominal or flank pain that can be diffuse or localized.***
- ***Spontaneous arterial rupture is most likely to occur in a person's twenties or thirties, but can occur at any point in life.***
- ***Cerebral arterial rupture may present with altered mental status and be mistaken for drug overdose.***
- ***Mid-size arteries are commonly involved.***

Arterial, intestinal, or uterine fragility or rupture usually arise in EDS Vascular type, but should be investigated for *any* EDS type. *Specialized information about EDS Vascular type and the effects of EDS on the vascular system can be downloaded or viewed on-line at ednf.org/medical.*

EHLERS-DANLOS NATIONAL FOUNDATION

This ***Medical Resource Guide*** brings you highlights of information found at **ednf.org**, where you can find resources about EDS available whenever and however you need them, from a quick summary for an anxious patient waiting in your office to in-depth articles and media.

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