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Connective Tissue Disorders Recognizing and Treating Structural Defects

Our bodies are held together by structures called connective tissue. Connective tissues make up the ligaments and tendons that help your joints move. They also provide structure and flexibility for body parts, such as your skin and blood vessels.

Damage to your connective tissue can cause serious health issues. Normal wear and tear from aging can start to erode connective tissues. So can certain autoimmune diseases, like lupus, and specific connective tissue disorders. The effects of these conditions can range from mild to severe.



Connecting the Dots

Connective tissue disorders aren't common. Several can be passed down genetically. Those tend to run in families. Each disorder affects different body parts.

One condition, known as Marfan syndrome, can lead to balloon-like bulges called aneurysms in blood vessels. These bulges are especially dangerous when they occur in the body's largest blood vessel, the aorta, which carries blood to the heart. If it bursts, it can cause sudden death.

Another disorder, called hyper-mobile Ehlers-Danlos Syndrome (or hEDS), can cause pain in the joints and other parts of the body. The condition epidermolysis bullosa (EB) weakens the skin. And the disorder osteogenesis imperfecta can make bones prone to breaking.

Although the symptoms and affected body parts are different, these conditions all stem from damaged connective tissues. The severity of symptoms for these disorders varies widely among patients. They can range from hardly noticeable to life-threatening (see Wise Choices box).

"Milder forms of these conditions are probably under-recognized and underdiagnosed," says Dr. Anna Bruckner, a skin expert at the University of Colorado.

The symptoms caused by connective tissue disorders can also often arise from many other health conditions. "And symptoms like chronic pain and chronic fatigue are often missed, or dismissed," explains Dr. Cortney Gensemer, a genetics researcher at the Medical University of South Carolina (MUSC), who has hEDS herself.

Tell your doctor if you have a family history of a connective tissue disorder or if you have a relative who died suddenly at a young age. That information can help with a diagnosis, explains Dr. Jason Cook, a vascular surgeon at the University of Nebraska Medical Center.

Finding the Cause

Sometimes, having a connective tissue disorder in the family can delay diagnosis, Gensemer explains.

"People can get accustomed to thinking that something painful is normal," she says. "You might say something to your mom and dad, and their answer might be 'Oh, that happens to me all the time.'"

But you can't always be sure what's causing your symptoms. So it's important to talk with your doctor about any symptoms you have. Your doctor will ask about your personal and family medical history. Some common tests that look for connective disorders include imaging tests or genetic testing.

Genetic testing on its own isn't enough to rule out some conditions, including Marfan syndrome, Cook says. That's because the test doesn't detect all possible genetic factors that can lead to the condition.

Researchers are continuing to search for the causes of connective tissue disorders. Gensemer, working with Dr. Russell Norris from MUSC, has built a registry of families living with hEDS. They're now carrying out a

large study looking for genes that can cause the condition.

“Having a genetic test would provide a definitive, easy, objective diagnosis,” Gensemer says. “It would also let people get a diagnosis earlier.”

Living With Connective Tissue Disorders

Researchers hope that a better understanding of the causes of these disorders will eventually lead to a cure. Currently, treatments for connective tissue disorders focus on managing symptoms.

Both Marfan syndrome and hEDS can affect your blood vessels, which makes them vulnerable to tearing. People with these conditions often need frequent monitoring. Some may eventually need surgery to make sure a blood vessel doesn't tear.

The same gene changes that affect the blood vessels in Marfan syndrome can also weaken the heart, Cook says. His team is studying how to repair blood vessels without putting extra stress on heart tissue.

Treatment for connective tissue disorders needs to be personalized, Cook explains, because the same condition can affect people very differently. Medications are available to help with some symptoms of these disorders, like high blood pressure and reduced bone strength.

Recently, the first two medications to help heal skin damage in people with EB were approved by the U.S. Food and Drug Administration. “So now having a diagnosis can open the door to treatment for these patients,” Bruckner says.

She and her team are working on ways to repair skin damage for people with severe EB. They're taking stem cells from patients' skin, fixing the broken gene, and then returning repaired cells to the body. They're testing whether these cells can heal blisters that arise in people with EB and prevent new ones from forming.

“The skin is visible, so we can just look to see whether this works,” Bruckner explains. “If it does, then we have the potential to change the stem cells into other types of cells that could potentially heal the person's other organs.”

“It's potentially one treatment, and you're done for the rest of your life,” adds Dr. Anthony Oro, a stem cell researcher at Stanford University.

Oro and others are now working on a large NIH-funded project to make it easier to get stem cell therapies manufactured, so the treatment can be used for more patients. They're starting with skin repair for EB. But other research teams are already looking at how to repair other tissues like bone, muscle, and heart tissue.

“We hope this technology will be applicable to many different disorders,” Bruckner says.

Could You Have a Connective Tissue Disorder?

Common signs and symptoms:

Marfan Syndrome

- Arms, legs, fingers, and toes that are long compared to your body.
- A chest wall that caves in or sticks out.
- A curved spine.
- A long, narrow face.
- Heartbeats that skip, flutter, or feel too hard or fast.
- Vision problems, like blurry vision or extreme nearsightedness.

Hypermobile EDS

- Joints that stretch farther than normal (joint hypermobility).
- Joints that pop out of place (dislocations).
- Skin that is soft or velvety, highly elastic (stretchy), or bruises easily.
- Chronic pain.
- Feeling tired all the time (fatigue).

Epidermolysis Bullosa (EB) and Related Skin Conditions

- Skin that blisters easily.
- Blisters in the mouth.
- Rough, thickened, or missing fingernails or toenails.
- Changes in the color of the skin.

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Tired, Achy Eyes?

Finding Relief for Eye Discomfort

Are your eyes tired, dry, or achy? Many factors can contribute to these types of symptoms. But a big culprit can be intense use of your eyes. Spending too much time looking at screens and held devices, like smartphones, can strain your eyes. So can normal aging. What can you do to find relief?

One major cause of eye discomfort is not blinking enough. "When we focus on tasks like reading or computer work, our blink rate just plummets," says Dr. Chantal Cousineau-Krieger, an NIH ophthalmologist.

Not blinking enough can cause your eyes to become dry and uncomfortable. Certain people are more prone to eye dryness, too. This includes those over age 50, women, and people who wear contact lenses. Certain medications, like antihistamines, and health conditions can also add to eye dryness.

Avoiding other factors that increase eye dryness may help your eyes feel better, too. Air blowing directly in your face from a fan or from air vents in the car can contribute to eye dryness, says Cousineau-Krieger. So can smoke or windy conditions.

Normal aging can also lead to eye strain. With age, we start to lose our ability to focus on close objects. This is called presbyopia. Our eyes need to work harder to focus.

“When we look at something up close, we flex the muscle inside of our eye,” Cousineau-Krieger explains. “And just like any other muscle, if you hold the contraction for a long time, the muscle can become fatigued. Eventually, in your 40s, you end up not being able to see things up close as well. It’s a natural part of aging that goes along with gray hairs and wrinkles. And then we typically need reading glasses to be able to see things up close.”

But eye strain doesn’t only happen to adults. Children can also develop symptoms from intensely using their eyes. They may not tell you that their eyes hurt. Instead, they may start blinking forcefully or rubbing their eyes.

Spending too much time on screens is also now believed to be contributing to children developing nearsightedness. Studies have shown growing rates of nearsightedness in children over the past few decades.

To relieve eye discomfort, you can try some simple steps. Experts recommend the 20-20-20 rule. Take eye breaks every 20 minutes and look far in the distance, about 20 feet away, for about 20 seconds.

“Experts are recommending that children spend time outdoors playing to help them focus on things further at a distance,” says Cousineau-Krieger. “Hopefully, this will also help decrease the amount of nearsightedness. The amount of nearsightedness is going up around the world.”

Taking screen breaks and focusing on more distant objects can be helpful for everyone’s eye health. If simple lifestyle changes don’t bring you relief from eye discomfort, it may be time to see a doctor for an eye exam.

Article reprinted from NIH-News In Health

Protect Your Eye Health

- **Rest your eyes.** Take a break from tasks that require focusing on nearby objects every 20 minutes, and look at something about 20 feet away for 20 seconds.
- **Limit screen time and take breaks often.** Encourage kids to spend more time outdoors and focus on things in the distance.
- **Keep your eyes moist.** Use eye drops called artificial tears when your eyes feel dry.
- **Prevent the air in your home from getting too dry.** Use a humidifier. Limit air conditioning.
- **Wear sunglasses when outside.** Choose sunglasses that block 99 to 100 percent of both UVA and UVB radiation.
- **Keep your eyeglasses and/or contacts prescription current.** Learn how to properly care for your contacts.
- **Maintain a healthy lifestyle.** Drink plenty of water, eat healthy, and get enough physical activity. Healthy habits can lower your risk for diseases and conditions that lead to eye or vision problems.
- **Quit smoking and keep your home smoke-free.** Get free help at smokefree.gov, 1-800-QUIT-NOW (1-800-784-8669), or by texting QUIT to 47848.



NOTICE OF ANNUAL MEETING OF MEMBERS

The Annual Meeting of the Members of the National Consumer's Advantage Association will be held at 12444 Powerscourt Drive, Suite 500A, St. Louis, Missouri, on Tuesday, December 17, 2024 at 9:30 a.m. (CST) for election of Directors and for the transaction of such other business as may properly come before the meeting and any adjournment thereof.

The above notice is given pursuant to the By-Laws of the Association.

PROXY

National Consumer's Advantage Association December 17, 2024 Annual Meeting of Members THIS PROXY IS SOLICITED ON BEHALF OF NATIONAL CONSUMER'S ADVANTAGE ASSOCIATION

The undersigned member of the National Consumer's Advantage Association does hereby constitute and appoint the President of National Consumer's Advantage Association, the true and lawful attorney(s) of the undersigned with full power of substitution, to appear and act as the proxy or proxies of the undersigned at the Annual Meeting of the Members of National Consumer's Advantage Association and at any and all adjournments thereof, and to vote for and in the name, place and stead of the undersigned, as fully as the undersigned might or could do if personally present, as set forth below:

1. FOR [], or to [] WITHHOLD AUTHORITY to vote for, the following nominees for Board of Directors:
Ben Malone, Joshua D. Farrell, and Daniel Malloy
1. In their discretion, the proxies are authorized to vote upon such other business as may properly come before the Meeting.

This proxy, when properly executed, will be voted in the manner directed by the undersigned member. If no direction is made, this proxy will be voted for the election of directors and officers.

Dated: _____, 2024

Signature _____

Name (please print) _____

Please date and sign and return promptly to NCA Association, 12444 Powerscourt Drive, Suite 500A, St. Louis, Missouri 63131 whether or not you expect to attend this meeting. The Proxy is revocable and will not affect your right to vote in person in the event that you attend the meeting.

St. Louis, Missouri
November 8, 2024
Date



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For information regarding your membership
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