

NIH News in Health

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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.

Researchers are looking for better ways to diagnose and treat conditions that lead to connective tissue damage. Getting treatment may help improve a person's quality of life. But connective tissue disorders can be hard to recognize.

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 hypermobile 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

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Definitions

Autoimmune Diseases

Conditions in which the body's disease fighting system mistakenly attacks and destroys the body's own cells.

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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

Definitions

Stem Cells

Cells that have the ability develop into many different cell types in the body.

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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. ■



Wise Choices

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.



Web Links

For more about connective tissue disorders, see "Find More Information" in the online article: newsinhealth.nih.gov/2024/09/connective-tissue-disorders

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. See the Wise Choices box for more eye health tips. If simple lifestyle changes don’t bring you relief from eye discomfort, it may be time to see a doctor for an eye exam. ■



Wise Choices

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.** To learn how to properly care for your contacts, see go.nih.gov/NIHNiH-Sep24-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.



Web Links

For more about eye discomfort, see “Find More Information” in the online article: newsinhealth.nih.gov/2024/09/tired-achy-eyes



Health Capsules

For links to more information, please visit our website and see these stories online.

How Aura May Trigger Migraine Pain

Migraine headaches can last for days. Some people have what's called an aura before migraine pain sets in. Aura can include visual disturbances, such as seeing flashing lights. And it can include other sensory changes, like numbness or tingling.

Scientists have known that aura is caused by a disruption of electrical activity within the brain. But they hadn't yet figured out how the aura might trigger pain. The nerve cells that drive migraine pain are known to sit outside the brain. And communication between these

nerves and the brain was thought to be blocked by a structure called the blood-brain barrier.

To learn more, researchers looked at how spinal fluid flows out of the brains of mice. They found gaps in the blood-brain barrier around a bundle of nerve cells known to process migraine pain. Substances injected directly into the brain flowed into these nerve cells within half an hour. This time period is similar to the typical time between aura and headache.

The scientists next provoked

migraine aura in the brains of mice. Then they measured changes in proteins that flowed into the nerve cells. They found changes in many proteins known to be involved in migraine headache. The results point to potential new ways to relieve migraine pain.

"These findings provide us with a host of new targets to prevent and treat migraines and strengthen existing therapies," says Dr. Maiken Nedergaard of the University of Rochester, who helped lead the study. ■

Don't Give In to Gum Disease

Periodontal, or gum, disease is an infection of the tissues that hold your teeth in place. It usually arises because of poor brushing and flossing habits. This can lead to a sticky film of bacteria called plaque, which builds up and hardens on teeth. The hard buildup, called tartar, can only be removed by a dentist or dental hygienist.

If not treated, gum disease can spread to the bones around the gums. It can make chewing painful. Eventually, it can lead to loose or lost teeth.

People with gum disease often have swollen, red, or bleeding gums. Other symptoms include persistent bad breath or sensitive teeth. People who smoke are especially at risk for gum disease. Other factors that raise your risk include hormone changes in girls and women, certain genes, and health conditions like diabetes and AIDS and their medications.

Treatment options range from non-surgical to surgical interventions. It's important to keep brushing and flossing every day. Changing

unhealthy habits and quitting smoking can be part of a treatment plan. Some patients may be referred to a specialist called a periodontist. These are doctors who can provide advanced treatment for gum disease.

To keep your teeth and gums healthy, use fluoride toothpaste and brush your teeth twice a day. Floss regularly or use another dentist-recommended device to clean between teeth. Be sure to visit your dentist regularly. Learn more at go.nih.gov/NIHNIHSep24Gums. ■



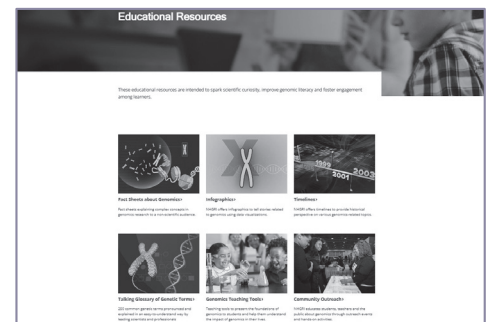
Featured Website

Genomics Educational Resources

www.genome.gov/about-genomics/educational-resources

Genomics is the study of all of the genes and DNA in a person or living thing. Learning about how your genome works can help you better understand your own

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