

WINTER 2020

NIH MedlinePlus

MAGAZINE

Trusted Health
Information from the
National Institutes
of Health

IN THIS ISSUE

Acid Reflux, Heartburn,
and GERD: What's the
Difference?

New Treatment
Options for Vitiligo

A Mother-Daughter
Journey with
Endometriosis

PLUS

Tips for Cancer
Survivors

Recognizing National
Donor Day

COVER STORY

Recording artist and
Broadway actress
Jordin Sparks shares
her family experience with

SICKLE CELL DISEASE

In this issue

Welcome to the first NIH MedlinePlus magazine issue of 2020!

We're excited to ring in the new year—and new decade—by sharing updates on key National Institutes of Health (NIH) research and health news with you.

In this issue, we take a look at sickle cell disease, a painful red blood cell condition that affects 100,000 people in the U.S., particularly African Americans and Hispanic Americans.

Multiplatinum recording artist and Broadway actress Jordin Sparks talks to us about the disease's impact on her family. She explains how losing her stepsister led her to advocate for others, especially those who are afraid to talk openly about the condition.

We also share a research update from NIH's sickle cell branch in our interview with Griffin Rodgers, M.D., who is also the director of the National Institute of Diabetes and Digestive and Kidney Diseases, and John Tisdale, M.D., of the National Heart, Lung, and Blood Institute. They discuss key advances in bone marrow treatment



John Tisdale, M.D. (left), and Griffin Rodgers, M.D., oversee sickle cell disease research at NIH.

and talk about next steps in research.

Other articles include: updates on long-term, breakthrough treatment options for vitiligo, an autoimmune disease that causes skin to lose color in patches; tips for cancer survivors as they transition back to a “new normal” after treatment; and additional features on heartburn and endometriosis, two common conditions that often go undiagnosed.

NIH MedlinePlus MAGAZINE

WHO WE ARE

The National Institutes of Health (NIH) is the **nation's premier medical research agency**, with 27 different institutes and centers. The National Library of Medicine (NLM) at NIH is a leader in research in biomedical informatics and data science research and the world's largest medical library.

NLM provides **free, trusted health information** to you at medlineplus.gov and in this magazine. Visit us at magazine.medlineplus.gov

Thanks for reading!

CONTACT US

Email
NLMScommunications@nlm.nih.gov

Phone
508-907-7000

CONNECT WITH US

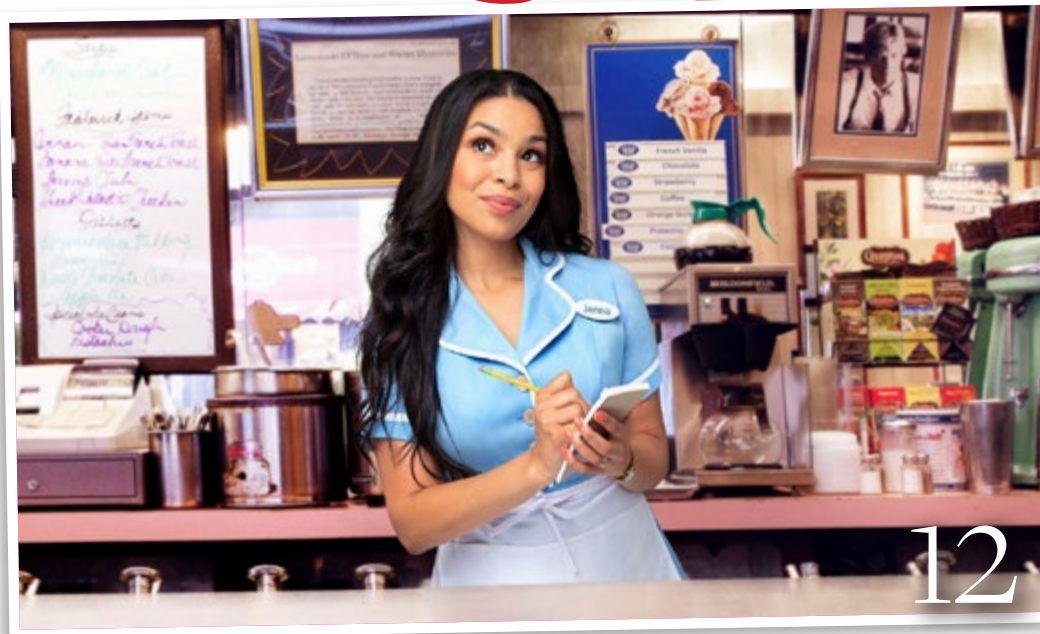
Follow us on Facebook
www.facebook.com/nationallibraryofmedicine/

Follow us on Twitter
[@NLM_News](https://twitter.com/NLM_News)



NIH MedlinePlus magazine is published by the National Library of Medicine (NLM) in conjunction with the Friends of the NLM.

Articles in this publication are written by professional journalists. All scientific and medical information is reviewed for accuracy by representatives of the National Institutes of Health. However, personal decisions regarding health, finance, exercise, and other matters should be made only after consultation with the reader's physician or professional advisor. Opinions expressed herein are not necessarily those of the National Library of Medicine.



“American Idol” winner and actress Jordin Sparks, pictured on the set of “Waitress” on Broadway, shares her late stepsister’s experience with sickle cell disease.

FEATURES

08 Heartburn: What you need to know

How to manage symptoms on your own and with your health care provider

12 Why Jordin Sparks wants more people to talk about sickle cell disease

Singer discusses removing stigma and honoring her late stepsister

18 Improving endometriosis diagnosis through research and awareness

Studies aim to find easier, less expensive options

24 Finding self-confidence with vitiligo

Patient shares the power of acceptance with others

DEPARTMENTS

04 To your health

News, notes, & tips from NIH

28 From the lab

Latest research updates from NIH

30 NIH on the web

Find it all in one place!

31 Contact us

NIH is here to help



Tips for
cancer
survivors

6



health

NEWS,
NOTES,
& TIPS
FROM NIH

What happens when your thyroid doesn't work correctly?

Hypothyroidism is the most common thyroid disorder

HEALTH TIPS January is National Thyroid Awareness Month, which brings attention to the small, butterfly-shaped gland in your neck.

The thyroid does a lot for your body. It makes hormones that keep organs running properly. It also controls the way you use energy: for example, how you burn calories and how fast your heart beats.

But sometimes this powerful gland doesn't work like it's supposed to.

One common thyroid condition, called hyperthyroidism, causes the thyroid to make more hormones than the body needs.

But the most common thyroid disorder is hypothyroidism, which affects about 4.6% of people in the U.S. This condition causes a lack of hormone, which leads the thyroid to slow down. People with hypothyroidism often gain weight and feel tired, but some may not have any symptoms at all.

Hashimoto's disease and hypothyroidism

The most common cause of hypothyroidism is Hashimoto's disease. It's an autoimmune disorder, meaning that the body's own immune system attacks the thyroid and causes inflammation. This prevents the thyroid from making enough hormones, causing hypothyroidism.

Hashimoto's disease often runs in families, and it affects women more than men. People with other autoimmune diseases are also more likely to get Hashimoto's disease.



Other causes

Other causes of hypothyroidism include thyroid surgery, radiation treatment, some medications, and thyroiditis (inflammation of the thyroid). Some people may be born with hypothyroidism, which is known as congenital hypothyroidism.

Women are more likely to have hypothyroidism, as well as people older than 60 and people with other thyroid disorders.

IMAGE: ADOBE STOCK

Symptoms

A person with mild hypothyroidism might feel fine and have no symptoms. But those with symptoms can experience a puffy face, sluggishness, weight gain, feeling cold, a slowed heart rate, constipation, depression, and thinning hair. Not everyone has the same symptoms, so make sure to talk with your health care provider if you think you have hypothyroidism.

Diagnosis

A doctor can diagnose hypothyroidism and help people manage it.

The doctor will ask about your symptoms, do a physical exam, and order blood tests. These tests measure your TSH, or thyroid stimulating hormone, level and antibodies. In Hashimoto's disease, high levels of antibodies in the blood show that the thyroid is being attacked by the immune system.

If you have an enlarged thyroid that's causing hypothyroidism, called a goiter, your doctor may order an ultrasound to see it better. In rare cases, you might need surgery, since an enlarged thyroid can make it hard to swallow.

Treatment

Hypothyroidism can usually be treated with an oral medication called levothyroxine. Levothyroxine replaces a hormone naturally produced by the thyroid gland to regulate the body's energy.

If your doctor prescribes levothyroxine, be sure to take it at the same time each day.

Your doctor may need to test the level of TSH in your body a few times to make sure the medication is working properly. TSH levels can change more during pregnancy, if you have heart disease, or if you take certain hormone therapies. ■

SOURCES: MedlinePlus; National Institute of Diabetes and Digestive and Kidney Diseases

National Donor Day is February 14

More than 113,000 people in the U.S. are waiting for organs

BY THE NUMBERS This Valentine's Day, share the love and learn about organ donation.

Every year on February 14, National Donor Day raises awareness about the lifesaving benefits of organ, eye, and tissue donation.

It also celebrates those who have saved lives through the gift of donation.

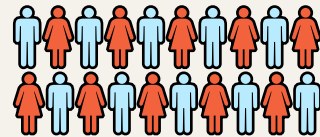
Organ donation takes the healthy organs and tissues of someone who has died and transplants them to people waiting for lifesaving organs. There are no age limits on who can donate, and even someone with an illness may be able to donate organs or tissue.

If you want to become an organ donor or learn more about it, visit the national organ donor website at <https://www.organdonor.gov>



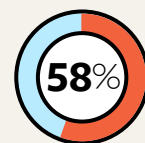
113,000 + men, women, and children were on the national transplant waiting list in 2019.

20 people die each day waiting for a transplant.



1 donor can save 8 lives by making vital donations of a heart, lungs, liver, pancreas, kidneys, and intestines.

95% of U.S. adults support organ donation but only **58%** are signed up as donors.



SOURCES: MedlinePlus; Health Resources and Services Administration



Life after cancer: Tips for finding your new normal

Strategies for survivors from survivors

HEALTH TIPS At the end of treatment, many cancer survivors have mixed emotions: grateful for treatment but happy to be done; excited to move on, yet scared, worried, or anxious about the future.

Some people want to return to the life they were living the moment before their diagnosis; others reassess what's important and what's next.

The National Cancer Institute (NCI) provides guidance for coping with life after cancer and staying healthy—both physically and mentally. Below are a few key takeaways to help survivors and their loved ones prepare for the next stage.

Talk to your health care team

It's likely that you already have a follow-up care plan, but if not, make sure to ask for one. There are also some additional things you and your care team can do.

Whether you're worried about symptoms you're experiencing or are anxious about your cancer returning, speak up. Your health care team can give you the

facts about your type of cancer and the chances of it returning. Just talking about your symptoms may help calm your fears.

Also make sure to write down your questions and anxieties as they come up. This can help you manage your concerns and provide a useful list to help you manage your next follow-up.

However, if thoughts about cancer returning start to interfere with your daily life, you may want to reach out to a counselor or therapist who specializes in treating cancer survivors.

Take care of your mind and body

Eddie Harris, who was diagnosed with testicular cancer at just 25 years old, says his number one suggestion is to surround yourself with a good group of people.

"It was important for me to let those in my inner circle know what I was thinking and feeling in those first few months following treatments—and to tell them what I needed," Eddie says.

In addition to your family and friends, online and in-person support groups can offer an additional kind of help.

Limiting stress is also key for survivors. Relaxation exercises, meditation, and yoga can all help with stress management.

Amy Fenske, who is a survivor of stage IV melanoma, tries to stay in the moment to help manage her stress.

“Nothing has forced me into the present more than cancer,” Amy says. “[My diagnosis] was a huge wake-up call to what’s important.”

Additionally, moderate exercise such as walking, biking, or swimming is a good way to feel better mentally and rebuild strength and endurance. Be sure to first check with your provider before starting or stopping any exercise routine.

Giving back

Volunteering in your local community or even helping other cancer survivors or patients can be a good way to give back and feel connected. Some activities include joining or leading a support group, contributing to an advocacy effort, or participating in a clinical trial.

Before giving back, make sure you feel emotionally ready and are not pushing yourself.

Food for thought: What you eat matters

After your treatment, eating well can help you regain strength, rebuild tissue, and feel better overall.

Here are some tips for healthy eating:

- Work with a dietitian to create a nutritious, balanced eating plan.
- Eat a variety of foods from all of the food groups.
- Choose high-fiber foods, such as whole-grain breads.
- Limit red meats.
- Limit alcohol to no more than one (for women) or two (for men) drinks per day.

For more information, check out NCI’s resources for cancer survivors at cancer.gov/about-cancer/coping/survivorship ■

SOURCES: MedlinePlus; National Cancer Institute

New on MedlinePlus: HIV PrEP and PEP

Medications work to prevent HIV

NEW ON MEDLINEPLUS Information about pre-exposure prophylaxis (PrEP) and post-exposure prophylaxis (PEP) is now available on MedlinePlus in both English and Spanish.

PrEP and PEP are medicines that work to prevent HIV, or human immunodeficiency virus. HIV is a serious condition that weakens your immune system, which can make you more likely to get infections and certain cancers.

In the U.S., HIV is most commonly transmitted through sex or contact with blood. The final stage of HIV is AIDS, or acquired immunodeficiency syndrome.

PrEP is a medication that people who are at high risk of HIV can take daily. PEP is for emergency situations, and should be taken within 72 hours of suspected exposure and for 28 days after.

You should contact your health care provider or go to



an emergency room right away if you think you may have been exposed to HIV. ■

SOURCE: MedlinePlus

Heartburn:

What you need to know



Hearthburn is a burning feeling in your chest or throat. Despite its name, heartburn actually has nothing to do with your heart.

Who gets it?

More than 60 million Americans experience heartburn at least once a month. Some studies suggest that more than 15 million Americans experience heartburn symptoms each day.

What causes it?

There are several conditions that can cause heartburn, but most often it's caused by acid reflux or a more chronic form of acid reflux called GERD, or gastroesophageal reflux disease.

Eating certain foods, drinking alcohol, and some medications can make heartburn more likely.

What is acid reflux?

When you swallow food, it passes from your throat, through your esophagus, and then through an opening in your stomach.

This opening usually closes once the food passes through. If it doesn't, acid from your stomach may enter through the opening and travel the wrong way—back into your esophagus and to your throat. This is called acid reflux. The opening can occur sporadically after a meal leading to multiple reflux events.

Is heartburn serious?

Occasional heartburn isn't a cause for concern. But if heartburn occurs regularly, interferes with your daily routine, or is caused by GERD, it could damage your esophagus or lead to cancer.

When heartburn is frequent or it leads to complications, it is categorized as GERD.

What are the symptoms?

- Burning in the chest after eating or at night
- Pain that worsens when lying down or bending over
- Bitter or acidic taste in the mouth

What makes heartburn worse?

Being pregnant or overweight can make you more likely to have heartburn. There are also certain foods and drinks that can trigger heartburn including:

- Spicy foods
- Acidic foods, like those with citrus (oranges or lemons) or those with tomatoes (marinara sauce)
- Alcohol
- Caffeinated beverages, like coffee and tea

FAST FACTS

More than **60 million Americans** experience heartburn at least **once a month**.

SOURCE: American College of Gastroenterology

How is it diagnosed?

If you're experiencing heartburn, make sure to track your symptoms. For example, how often does it happen in a day? Does it happen after eating? You can bring this list to your provider to help make a faster and better diagnosis.

After discussing your symptoms and medical history with you, your provider can usually diagnose heartburn easily.

What are the treatment options?

If you have heartburn, your provider may prescribe over-the-counter antacids, which may come in a chewable tablet. They might also recommend prescription drugs such as H₂ blockers or proton pump inhibitors, which reduce how much acid your stomach makes.

Providers may also recommend lifestyle changes, like losing weight and quitting smoking, which can increase stomach acid production. Other changes often include reducing stress, exercising more, avoiding food around bedtime, raising the head of the bed about 6 inches, and eating more high-protein, low-fat meals.

What if medication or lifestyle changes don't help?

Your provider may request more testing if lifestyle changes and medication did not help. These tests include a pH test to check for acid in your esophagus or an endoscopy. This test uses a small tube with a camera and light to see inside of your digestive tract and spot any issues. ■

SOURCES: MedlinePlus; National Institute of Diabetes and Digestive and Kidney Diseases; American College of Gastroenterology

4 important heartburn facts you should know about

Dr. John Pandolfino talks triggers, weight loss, and complications

Despite what it's called, heartburn—or that burning feeling in your chest—has nothing to do with your heart. Here are four key facts about heartburn from John Pandolfino, M.D., of Northwestern University.

1 Many diseases can cause heartburn. Acid reflux is not always the cause. One disease that can cause heartburn is achalasia, a rare disorder that makes it difficult for food and liquid to pass into your stomach. Another is eosinophilic esophagitis. That's an allergic inflammatory disease of the esophagus.

3 Not all foods trigger heartburn. There's a difference between what makes you more likely to experience heartburn and what triggers heartburn. Fatty foods, for example, lower your anti-reflux barrier, which can increase your chances of heartburn. Acidic foods may trigger heartburn the moment they hit your esophagus. Avoiding heartburn triggers will help stop heartburn.

2 Regular heartburn does not lead to cancer. Most people who experience occasional, mild heartburn will not develop esophageal cancer. If you have heartburn regularly for 10 or 15 years, for example, there is a very small chance that it could lead to cancer. Always talk to your doctor if you experience regular symptoms and please don't wait, as endoscopy can assess your risk.

4 Significant weight loss is not necessary to ease heartburn. Some people experience heartburn after gaining weight. However, significant weight loss—such as 50 or 60 pounds— isn't necessary to stop heartburn. Losing the 5 or 10 pounds you gained and returning to your baseline weight will help heartburn disappear.

SOURCES: MedlinePlus; National Institute of Diabetes and Digestive and Kidney Diseases

What's new in heartburn research?

Imaging advances help doctors better understand long-term treatment needs

Working with NIH's National Institute of Diabetes and Digestive and Kidney Diseases, John Pandolfino, M.D., studies how to improve intestinal pain like heartburn.

He works at Northwestern University, where he teaches and leads a team of researchers.

Dr. Pandolfino spoke to NIH MedlinePlus magazine about new research in this area and advice for people experiencing heartburn and related issues.

Why did you decide to study heartburn?

When I was a fellow and trainee at Northwestern University, some of the best research focused on gastrointestinal motility and physiology. This refers to the contraction of the smooth muscles in the gastrointestinal tract.

What inspired me about this area is how logical it is. It uses laws of fluid dynamics and biomechanics.

It was also interesting to me because I come from a family of plumbers. In some ways, I feel like I stayed in the family business. I'm just a human plumber that tries to prevent clogs, blockages, and things from going backwards.

Tell us about your recent research.

We've been looking at how well diagnostic tests can identify specific markers and measurements that may help us predict who needs to reduce or increase their reflux therapy.

For example, we use a wireless capsule that monitors the overall acid burden over four days. You swallow it, and it safely travels through your digestive tract just like a piece of food.

This can help us predict who with heartburn symptoms actually needs a PPI—or proton pump inhibitor. PPIs are a group of drugs whose main purpose is to reduce the production of stomach acid. We want to identify which patients can get off it in favor of other therapies and those who truly need it.

What has your research found?

We can now help patients better understand why we have to keep them on medicine—or don't.

For example, for people who do not show any evidence of acid burden through capsule imaging, they can stop their PPIs.

We also found that in people who do have considerable acid burden, we could more easily identify who might need something more dramatic than PPIs, such as an endoscopic or surgical procedure to strengthen the reflux barrier.

What effects might this have on how health professionals treat patients?

It helps give patients an important message that we're not guessing whether or not you need this medicine—we're telling you. We're not going to give you medicine if it's not going to help you.

It sounds very simplistic, but this would be such a cost-effective strategy for people.



“For people on PPIs, never look at it as if you have to have this kind of treatment for the rest of your life.”

– John Pandolfino, M.D.

John Pandolfino, M.D., is trying to improve treatment for people experiencing heartburn.

What advice do you have for people experiencing heartburn?

See your primary care provider. They will assess your overall risk for developing something called Barrett's esophagus. This occurs when the cells in the lining of your esophagus are damaged from acid reflux. If the cells develop abnormally, it could lead to esophageal cancer.

People with heartburn can do plenty to manage symptoms on their own, too. This includes eating well, losing weight, exercising, and reducing stress. Stress reduction will cause a dramatic reduction in your symptom severity.

For people on PPIs, never look at it as if you have to have this kind of treatment for the rest of your life. If you have heartburn and need a PPI, you should first work on lifestyle modifications. It's amazing how many people can get off the PPI and maybe just use antacids when they need them. ■

Finding her voice: A teacher's heartburn experience

Laurie Hayes never expected acid reflux was causing her hoarseness



Acid reflux treatment helped teacher Laurie Hayes improve her vocal issues.

For nearly 20 years, Laurie Hayes struggled with losing her voice.

The preschool teacher, who once sang in local bands, never imagined that a condition that affects nearly 60 million Americans would be to blame: heartburn.

"Doctors first diagnosed me with vocal cord nodules, which made it difficult for me to speak," Laurie says. "I had seen speech pathologists and other doctors for years, and we decided it was best to have surgery to remove them."

Vocal cord nodules aren't dangerous, but they can cause hoarseness and discomfort.

Getting answers

In 2010, Laurie met with a specialist to address her hoarseness.

He ordered a laryngoscopy, a procedure that lets a doctor look at your throat and voice box. Following this procedure, they scheduled the surgery, which required not speaking for a month leading up to it.

During that period, Laurie's doctor prescribed a medication that treats acid reflux and heartburn, she says.

Acid reflux occurs when the contents of your stomach back up into your esophagus.

PERSONAL STORY Heartburn is a burning sensation behind your breastbone, neck, or throat that is often caused by acid reflux.

"When I started taking the medications he prescribed, I was shocked that my voice started coming back," said Laurie.

Not only did her voice return while taking the medication, but she realized that the pain she had been feeling was not her injured vocal cords, but heartburn.

"I didn't realize how much it was hurting my throat until I missed a dose," she says. "I would get that burning feeling in my throat and my voice would go away. When I took my medication, I started feeling a lot better and my voice would come back."

Laurie's voice improved so much with the use of the acid reflux medication that she canceled the nodule surgery.

"Suddenly I had my life back—and the fact that I had my voice back was so crazy," she says.

Life managing heartburn

In the nine years since, Laurie has been on a heartburn medication to manage her symptoms and meets with her doctor regularly.

She says heartburn will be something she has to manage forever and that while she experiences occasional hoarseness, her voice is back to normal.

Along the way, Laurie says she's learned a lot about managing her symptoms with lifestyle changes.

She avoids foods like tomato sauce or anything spicy late at night, both of which trigger her heartburn. She's also careful not to overeat and she avoids caffeinated drinks like coffee and soda.

"Suddenly I had my life back—and the fact that I had my voice back was so crazy."

– Laurie Hayes

When she's not so careful with her diet, she needs to take her medication to counteract the effects.

"My advice for others would be to pay attention to your symptoms and talk to your doctor," Laurie says. "Don't ignore anything because heartburn can really affect your body." ■

Find Out More

- ▶ **MedlinePlus: Heartburn**
<https://medlineplus.gov/heartburn.html>
- ▶ **MedlinePlus: GERD**
<https://medlineplus.gov/gerd.html>
- ▶ **Acid Reflux in Adults**
<https://www.niddk.nih.gov/health-information/digestive-diseases/acid-reflux-gerd-gerd-adults>
- ▶ **ClinicalTrials.gov: Heartburn**
<https://clinicaltrials.gov/ct2/results?cond=heartburn>

Why Jordin Sparks wants more people to talk about sickle cell disease

After losing her stepsister, singer works to remove stigma and give more patients a voice

Multiplatinum recording artist Jordin Sparks first got the nation's attention as the youngest winner of the hit TV singing competition show "American Idol" in 2007. Since her win, Sparks has written and produced Grammy-nominated albums and found an additional calling as an actress. Just last fall, she starred in the Broadway musical "Waitress."

Offstage, Sparks has become a leading advocate for people with sickle cell disease. She lost her stepsister, Bryanna, to the blood disorder last year. Since then, Sparks has brought more attention to the disease and started an open conversation with people with sickle cell disease and their loved ones. She shared her experience with NIH MedlinePlus magazine.

Tell us about your stepsister's journey with sickle cell.

Bryanna passed away in January of 2018. It was really hard. She was just 16 years old. She fought so hard her entire life.

But she always went through things—even when they were hard—with a smile on her face. She was just like any other kid. She loved music and fashion. She wanted to be a model. She was one of the kindest and strongest people I have ever known.

Brie's dream was to raise more awareness about sickle cell disease and to find a cure for it.

What did you know about sickle cell before you learned of her diagnosis?

I didn't know much. When my mom met my stepdad, I got to know Brie and then learned so much more about it.

I watched her struggle and saw how it affected her daily life. She had to miss school. Sometimes she was in the hospital for three weeks out of the month. Sometimes the pain was so bad she couldn't speak. Seeing Brie's experience, I had to get involved and use my voice to raise awareness about sickle cell disease and open the conversation.

What is your message to those with sickle cell?

It's important for you to know that you are not alone. There are people who know and understand what you are going through.

Being able to change the narrative about sickle cell disease is incredibly important because you can't tell that someone has it just by looking at them. We need to end the stigma that can come with sickle cell disease. No one should feel alienated because they are in pain.



This past fall, Sparks starred in the Broadway musical "Waitress."

That is why it has been amazing to be part of the Generation S storytelling project along with the Sickle Cell Disease Association of America and Novartis.* It's important for people to come together and share their sickle cell stories.

Why is research like that supported by the NIH and others so important?


Sickle cell disease is a rare disease that doesn't get enough attention. We need more research to find a cure.

What is the latest with your career as singer, songwriter, and actress?

I'm continuing with my music, adding more songs to the project I am working on. I was also recently onstage in the Broadway musical "Waitress." Finally, I'm working on a new Christmas-themed movie that I think Bryanna would have really loved.

I also want to continue my advocacy work with sickle cell disease to make a difference and spread awareness to help others. ■

** NIH was not involved in the creation of the project and reference to it does not constitute or imply endorsement by any federal agency.*



"We need to end the stigma that can come with sickle cell disease. No one should feel alienated because they are in pain."

– Jordin Sparks

Updates from NIH's sickle cell disease scientists

Dr. Griffin Rodgers and Dr. John Tisdale discuss bone marrow transplants, gene therapy

Many people with sickle cell disease (SCD) are healthier and living longer thanks in part to research led and supported by the National Institutes of Health (NIH). Recent efforts have led to breakthroughs in treating and even curing the red blood cell disease.

In order to have SCD, a person must inherit the sickle cell trait from their birth mother, father, or both parents. If you have just one trait, you can live a very long time. If you have the trait from both parents, SCD can be extremely painful and lead to death.

"Many people don't realize the severity of this disease," says John Tisdale, M.D., senior investigator at NIH's National Heart, Lung, and Blood Institute, who leads NIH sickle cell disease research along with Griffin Rodgers, M.D., director of the National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK).

"It can cause severe, debilitating pain that stops you from doing all the things in life that people try to accomplish," Dr. Tisdale adds. "It affects patients and families and support systems."

In the U.S., more than 100,000 people have the disease and it's more common in African Americans and Hispanics. Millions of people worldwide suffer from the disease too, including large populations in India and the Middle East.

The breakthroughs have been remarkable and lifesaving for many. However, not all those with SCD can be substantially helped or cured by current treatments. And many patients, especially those in other countries, have limited access to current treatments for the disease.

Researching more cures

That's why researchers like Dr. Rodgers and Dr. Tisdale continue testing new ways to treat the disease.

They've been working side by side for years with the same goal in mind: to cure *all* people in simpler, less expensive ways.

Their recent research aims to help more patients find a cure through expanded bone marrow transplant treatment and gene therapy.



John Tisdale, M.D. (left), and Griffin Rodgers, M.D., lead sickle cell disease research at NIH.

"If we could cure sickle cell disease in a safe and effective way, such as a pill that can reverse the disease, that would be a home run."

– John Tisdale, M.D.

"The majority of patients with SCD are still suffering as they were a decade ago despite these advances," says Dr. Rodgers.

Other treatments for SCD include hydroxyurea, a medication developed for this use by Dr. Rodgers and colleagues. It has been approved for use in adults for years but was only recently approved for children in the U.S.

While it's not a cure, hydroxyurea helps red blood cells better carry oxygen in patients with moderate to severe SCD. This lessens pain and helps patients live longer.

How bone marrow transplants work

In a more traditional bone marrow transplant, a patient is first treated with chemotherapy. Then, he or she receives a bone marrow transplant from a healthy donor.

But bone marrow transplants are often expensive, time consuming, and hard to get.

"One problem with bone marrow transplants is that you need to have a family member whose genes match," says Dr. Rodgers.

Other problems include the risk of stroke, which people with SCD are more susceptible to because of the disease's heavy stress on their organs.

More treatment options

To address these limitations, Dr. Rodgers, Dr. Tisdale, and their research teams are studying how to *repair* bone marrow through gene therapy. This treatment doesn't require a donor.

"Now we can take the patient's own bone marrow, use an engineered virus that carries genes to the bone marrow, and give repaired bone marrow stem cells back to the patient so that healthy bone marrow cells can grow," Dr. Tisdale says.

The procedure has been effective in more than 90% of adults in a number of small clinical trials already.

Researchers are working hard to test it in larger clinical trials soon.

Future of sickle cell treatment

Ultimately, Dr. Rodgers and Dr. Tisdale hope to work towards more affordable, accessible treatment options—such as bone marrow repair.

"If we could cure sickle cell disease in a safe and effective way, such as a pill that can reverse the disease, that would be a home run," says Dr. Tisdale.

Dr. Rodgers adds: "It sounds like science fiction now, but that's what we think about for the future."

For those with the disease, Dr. Rodgers and Dr. Tisdale recommend seeking out a clinical trial through ClinicalTrials.gov. Patients can get access to new treatment options and help researchers like Dr. Rodgers and Dr. Tisdale better understand the disease. ■

SOURCES: MedlinePlus; Centers for Disease Control and Prevention

Sickle cell disease: What you need to know

Symptoms, treatments, and causes

Sickle cell disease (SCD) or sickle cell anemia is a genetic blood disorder that causes red blood cells to change shape and to block blood vessels, causing severe pain. The blockage can lead to infections, organ damage, and stroke.

What causes it?

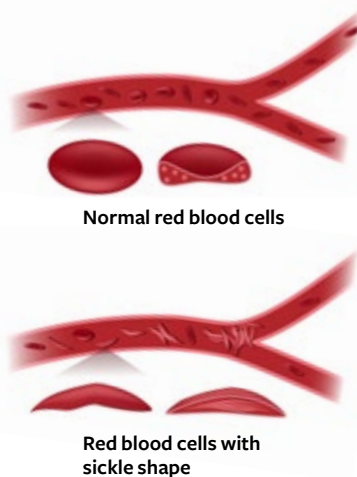
SCD is inherited from one or both parents. You cannot catch SCD from another person. SCD causes red blood cells to change shape from flexible smooth discs to hard, sticky "sickle" shapes. The sickle shaped cells can get stuck in small blood vessels and block the flow of oxygen to organs in your body.

Who is affected?

SCD affects 90,000 to 100,000 people in the U.S. It mostly affects people with African American, Hispanic, and Mediterranean heritage. Worldwide, SCD affects millions of people.

What are the symptoms?

- **Swelling:** Painful swelling in your hands or feet is one of the earliest signs.
- **Anemia:** A lack of red blood cells, or hemoglobin, can cause extreme tiredness and weakness.
- **Severe pain:** Sickle cells get clogged and block blood flow, which can be extremely painful. The pain cannot be predicted and can start suddenly.
- **Stroke:** In severe cases, sickle cells can clog blood flow to the brain and cause a stroke.
- **Eye disease:** SCD can affect the blood vessels in the eye and damage the eyes.
- **Acute Chest Syndrome:** Sickle cells can block blood flow to the lungs and chest, which makes it hard to breathe. These symptoms are similar to those of pneumonia, causing coughing, chest pain, difficulty breathing, and fever.



How is it treated?

SCD can be treated with medication including the antibiotic penicillin, blood transfusions, and blood or bone marrow transplants. The only cure for SCD is a bone marrow transplant. However, there can be complications with bone marrow transplants and not everyone can receive one. ■

SOURCES: MedlinePlus; National Heart, Lung, and Blood Institute; Centers for Disease Control and Prevention

From Africa to the U.S.:

A young woman's search for sickle cell disease treatment

Bone marrow transplant gives family hope



"I can't describe the pain," says Yinka.

"I had it everywhere in my body—my chest, my back, my face, and even in my fingernails."

Yinka is a 37-year-old second grade teacher in Baltimore, Maryland. When Yinka was only 1 year old, she was diagnosed with sickle cell disease (SCD), a painful genetic disease that causes problems with red blood cells.

Growing up, she knew she was different because of SCD.

"I wasn't like the other children in school," she says. "I was small. I couldn't run as fast." She also missed a lot of school due to doctor's appointments and illness.

"There was pain wherever the blood flows. It hurt so badly," she recalls.

PERSONAL STORY Yinka grew up in Nigeria, Africa, where access to new and developing treatments for the disease was limited.

Determined to get Yinka the help she needed, her mother moved their family to the U.S.

But in the U.S., Yinka continued to struggle. She remembers constantly going to her doctor, explaining the pain and looking for any kind of help.

"I would look at him in the eyes and say, 'Something is wrong.' But the doctor would just send me home each time with medication."

When she was in her early 20s, Yinka and her mother started exploring bone marrow transplants, the only cure for sickle cell disease to date.

"I remember watching a boy with sickle cell disease playing in the hospital waiting room. He looked so healthy, not small, not with yellow eyes, like me," Yinka recalls.

Her mother learned that the boy had received a bone marrow transplant from his younger brother. She wanted the same for her daughter.

A perfect match

Yinka was very fortunate to have a younger sister who was a half-match for a bone marrow transplant.

"My sister hates needles, but she took it all like a champ," Yinka says. "I knew she was doing it for me."

The transplant process was long. It involved months of drawing blood, stem-cell injections, hospitalizations, kidney monitoring, and much more.

But despite the lengthy and sometimes challenging process, Yinka's family finally got the news they had been waiting for.

"I woke up one morning, and something was different. I actually felt good. For the first time ever, I woke up without pain," Yinka says.

She and her mom were jumping for joy and laughing.

"You tell people you have pain, but they just don't get it. For the first time, I knew what it was like to not have any pain," Yinka adds.

Spreading awareness

When a person has sickle cell disease, they experience severe pain every day.

Yinka hopes to build awareness about this pain so that people take the disease and the patient seriously.

"It's a chronic disease and we aren't lying," Yinka says. "We need to take care of the severe pain that we are in." ■



Yinka, right, shares her sickle cell treatment story along with NIH researcher Courtney Fitzhugh, M.D.



Courtney Fitzhugh, M.D., is working to expand treatment options for sickle cell patients of all ages.

Sickle cell disease (SCD) specialists have performed bone marrow transplants in children since the 1990s. This procedure is the only cure for SCD to date.

NIH MedlinePlus magazine spoke with Courtney Fitzhugh, M.D., a Lasker Clinical Research Scholar at the National Heart, Lung, and Blood Institute, about the current and future outlook for this important treatment.

What NIH-supported SCD research are you working on?

Since the 1990s, to do a bone marrow transplant, a patient had to have a sibling with a complete tissue match and take a medication that suppresses their immune system (Cyclophosphamide), as well as high dose chemotherapy.

This has been highly successful in children. The problem is, most adults can't tolerate high doses of chemotherapy because their organs are already damaged and they don't have a sibling with an exact tissue match.

My research involves giving medications to suppress the immune system in adults. We are trying to create a state where instead of completely replacing bone marrow with that of their donors, we are creating a mixture of donor and patient cells because we now know that as low as 20% donor cells is enough to reverse sickle cell disease.

Investigating bone marrow transplants:

A cure for some sickle cell disease patients

NIH researcher works to improve transplant success for adults

If we use a donor who is half-matched, we can increase the chance that a patient will have a suitable donor from 10% to 90%. Parents and children can be a half-match.

What prompted you to do this research?

About 95% of children can be cured if they have a sibling with a complete tissue match. Adults with a sibling who have a tissue match can also survive with immune suppression medication. I wanted to develop a way to have half-match donors contribute bone marrow and for the outcomes to be successful.

What challenges do you face when researching SCD?

We had a lot of patients referred to us that we couldn't help because they didn't have a donor who was a tissue match. Then when we developed a half-matched protocol, half of the patients eventually rejected the cells, and the disease came back. So I wrote a new protocol in 2017 which has been much more successful so far.

What types of changes are you seeing?

Bone marrow transplants are not working in 100% of patients. I want to make changes to make them more successful. Also, I'd like to figure out why a transplant works in some patients rather than others and also focus our approach instead of generally suppressing the whole immune system.

I'm hoping our research can go beyond sickle cell disease to have a bigger impact.

What advice do you have for people with SCD?

Keep an open mind about new opportunities that are available. There are a lot of resources for care and treatment. Also, talk to your doctor and join a clinical trial in your area.

There's a lot of hope. It's an exciting time for sickle cell disease research. Not everyone can get a bone marrow transplant. However, there are a lot of studies, drug trials, and therapies. It's a different time now—it's a better time. ■

Find Out **More**

- ▶ **MedlinePlus: Sickle Cell Disease**
<https://medlineplus.gov/sicklecelldisease.html>
- ▶ **National Heart, Lung, and Blood Institute: Sickle Cell Disease**
<https://www.nhlbi.nih.gov/health-topics/sickle-cell-disease>
- ▶ **NIH News in Health: When Blood Cells Bend**
<https://newsinhealth.nih.gov/2012/04/when-blood-cells-bend>
- ▶ **ClinicalTrials.gov: Sickle Cell Disease**
<https://clinicaltrials.gov/ct2/results?cond=sickle+cell+disease>

Improving endometriosis diagnosis through research and awareness

Dr. Lisa Halvorson encourages women to speak up about their pain

Along with the growing awareness about endometriosis and how many women it affects, researchers have also been working to better understand the disease and its symptoms, so they can improve diagnosis and treatment—and ultimately find a cure.

NIH MedlinePlus magazine spoke with Lisa Halvorson, M.D., chief, Gynecological Health and Disease branch of the Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD). Dr. Halvorson discussed the latest endometriosis research and goals for future treatments.

What prompted NICHD researchers to do research around endometriosis?

NICHD has a strong interest in supporting research which improves women's reproductive health. Endometriosis is a common disease that is debilitating for many women and a substantial cause of infertility. We do not understand the causes of this disorder and have limited options for diagnosis and treatment. There is enthusiasm for pursuing all of the unanswered research questions since they may have a significant impact on women's lives.

What's the latest in National Institutes of Health (NIH)-supported endometriosis research?

NIH has supported work which has led to the development of new treatment options for endometriosis. One of these is the newly available oral medication, Elagolix, which has proven effective for treating endometriosis pain. However, this medication can have side effects and women on this medication cannot get pregnant. Therefore, additional studies are being supported to identify other treatment options.



Lisa Halvorson, M.D., wants to improve treatment and ultimately find a cure for women with endometriosis.

We urgently need more research to better understand the basic biology behind the development of endometriosis, including the role of genetics, stem cells, and immunology in the disease. This understanding could provide new ideas for treatment options.

We currently diagnose endometriosis with laparoscopic surgery, although there is a move towards basing the diagnosis on the presence of symptoms alone. It would be great if we could make an accurate diagnosis using a simple, inexpensive, non-invasive approach such as a blood test or a test of the menstrual tissue. This would be similar to the use of stool samples to screen for colon cancer. There is currently a lot of interest in developing a diagnostic tool for endometriosis that doesn't require surgery.

How has research contributed to our changing understanding of endometriosis?

Through research, we are realizing that endometriosis comes in many different forms and has a wider array of presentations than we previously appreciated. We have begun to understand that endometriosis is actually a systemic disease. That is, although the lesions are seen in the pelvis, there are widespread effects throughout the body including an increased risk of heart disease, depression, and some cancers. There is still more to learn but many of these associated disorders may be due to activation of the immune system.

DID YOU KNOW?

Endometriosis is especially common among women in their 30s and 40s and may make it harder to get pregnant.

SOURCE: Office on Women's Health

"We urgently need more research to better understand the basic biology behind the development of endometriosis."

- Lisa Halvorson, M.D.

What types of challenges do researchers face?

One of the big challenges is how to study a disease that appears in a wide variety of ways. Some women can have a huge amount of endometriosis—lots of tissue outside of where it should be—but little pain. Other women may not appear to have much endometriosis but have severe pain and infertility.

We also have difficulty diagnosing the disease. A lot of girls have it in adolescence but don't speak up until they are in their twenties. If you don't know the natural course of the disease, it's harder to understand what's driving it, what makes it worse, or what the symptoms are.

What is awareness of endometriosis like right now?

The good news is that there's more awareness among health care providers and women than there was just a few years ago. However, health care providers need to be more careful when taking patients' medical histories and patients need to feel more comfortable providing open and accurate answers. Instead of saying, "I'm just fine," women need to speak up and say, "Actually, my periods are just horrible."

What is your hope for the future of endometriosis research?

I'd also like to see ways to prevent rather than just treat the condition so that women don't suffer. That would be the holy grail. ■



Endometriosis: What you need to know

The uterus, or womb, is where a baby grows in a woman's body when she is pregnant. It's lined with tissue known as the endometrium, which is key for implantation, growth, and growth of the placenta. All of these are critical to a baby's health.

But if a woman isn't pregnant, that tissue sheds monthly along with menstrual blood, a process known as menstruation, or a period.

For women with endometriosis, tissue that is similar to the lining of the uterus grows in other places in their body, including:

- On the ovaries
- On the fallopian tubes, which carry eggs from the ovaries to the uterus
- Behind the uterus
- On the bowels or bladder

Just like the tissue that is shed from the uterus each month during a woman's period, the tissue in these other places can also shed, which could cause pain, bleeding, and possible complications.

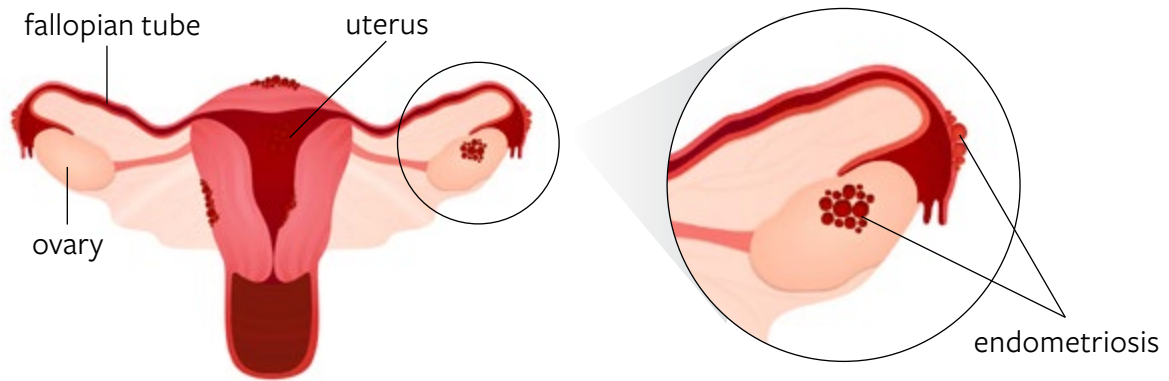
Who is affected?

Endometriosis can affect any woman who menstruates. In the U.S., it is estimated that 10% to 15% of women of childbearing age have endometriosis.

Surgery has long been considered the gold standard for confirming the diagnosis of endometriosis.

What are some of the risk factors?

- Periods started at an early age (before age 11)
- Periods are short or random, or last more than 7 days, and are heavy
- Abnormal vagina, uterus or fallopian tubes
- Having a mother, sister, or daughter with endometriosis



What are the symptoms?

The main symptoms of endometriosis are infertility and pain during your period. Although often worse during your period, pain and other symptoms can also occur throughout the month, including:

- Extreme lower abdominal pain or bloating
- Lower back pain
- Pain with bowel movements or during urination
- Nausea, vomiting, or feeling light-headed
- Heavy vaginal bleeding during your period or spotting in between periods
- Pain during sex
- Chronic fatigue

Women with endometriosis may have some or many of these symptoms. Be sure to discuss all of your symptoms with your health care provider to make sure you are diagnosed correctly.

How is it diagnosed?

Surgery has long been considered the gold standard for confirming the diagnosis of endometriosis. The most common type is called a laparoscopy, which uses a tiny camera attached to a thin tube called a laparoscope to see inside a woman’s reproductive areas and around her intestines. Sometimes a small sample of tissue, called a biopsy, is taken during surgery to confirm the diagnosis.

Doctors can also use imaging techniques, such as an ultrasound or magnetic resonance imaging (MRI), to help diagnose the condition.

How is it treated?

Endometriosis can be treated but not cured.

Medications that contain hormones, such as birth control pills, injections, or nasal spray can help lessen pain and bleeding. However, the symptoms can come back if the medicine is stopped. Providers may also prescribe pain medication.

Surgical treatment to remove small sections of scar tissue can be done using laparoscopy. A laparotomy is a more invasive procedure where a surgeon makes a cut into the abdomen to explore or remove scar tissue. Other surgical treatments include cutting nerves to help with pain.

If you suspect endometriosis, make sure to reach out to your doctor or gynecologist, a doctor who focuses on female reproductive organs. They will help review your medical history, discuss symptoms, as well as help with diagnosis and potential treatment. ■

SOURCES: MedlinePlus; Eunice Kennedy Shriver National Institute of Child Health and Human Development

60%–80%
of female patients
with chronic
pelvic pain have
endometriosis

Approximately
5 million
U.S. women's
lives are impacted
by endometriosis

Roughly
176 million
women worldwide
suffer from
endometriosis

Women with
endometriosis have an
increased risk
of ovarian cancer,
depression, and migraines

SOURCE: Eunice Kennedy Shriver National Institute of Child Health and Human Development

Inheriting endometriosis

A mother and daughter share their journeys and offer advice

A mother's perspective

"How do you measure pain when no one talks about it?"

That's what Suzanne Chutchian, a licensed clinical social worker and therapist, remembers about her endometriosis before she got treatment.

Endometriosis happens when tissue similar to the lining in a woman's uterus grows in other parts of the body. It can cause heavy bleeding and severe back and abdominal pain during a woman's menstrual cycle, or period. It can also cause intestinal problems and infertility.

Suzanne recalls trying to walk up the stairs at Boston University when she was 27 years old.

"I couldn't do it—the pain was horrible," she says. "My friend told me that menstrual cramps shouldn't prevent me from climbing stairs."

That's when Suzanne decided to discuss her symptoms with a gynecologist, a doctor who specializes in women's health.

A hereditary condition

Before getting help, Suzanne tried to manage her pain with over-the-counter pain relievers like aspirin and ibuprofen. She also used hot-water bottles to help ease her stomach cramps.

Her condition had a big impact on her daily life. In addition to painful menstrual cramps, she also had bad digestive pain and unpredictable bowel movements during her period.

Suzanne's mother also had endometriosis. But it wasn't something that was discussed much back then, so Suzanne, like many women, didn't know what to do about it.

"My mother thought it was somewhat normal to have pain, but she still encouraged me to seek treatment for it," she says.

Two options

When Suzanne finally discussed her pain and other challenges with her gynecologist, he gave her two options: laparoscopic surgery or pain killers.

She chose laparoscopic surgery, which confirmed that she indeed had endometriosis and treated it.

"After the surgery, I had no pain at all, and no stomach or bowel pain," she says. "I had freedom! I could do anything I wanted to do."

Pregnancy issues

Because endometriosis can produce scar tissue in the pelvis, it can be difficult for women to become pregnant. So Suzanne's doctor prescribed medication to help Suzanne increase her chances of getting pregnant.

After several years of trying to get pregnant with no success, Suzanne looked into adoption.

A year later, soon after she adopted a baby boy, her son Eric, she learned she was pregnant.

Her pregnancy was not difficult, but she had frequent bleeding. Twelve weeks into her pregnancy, Suzanne learned that she had been carrying twins, but only one baby survived.

That baby, her daughter, Rachel, is now 25 years old. Rachel also has endometriosis.

Paying it forward

Suzanne is glad that today there are more treatment options for Rachel and other women with endometriosis. Suzanne has had a number of laparoscopic surgeries since her first one. Doctors also removed the parts of her ovaries that were covered with excess tissue from endometriosis.

"If you have pain and other symptoms of endometriosis, don't downplay your symptoms."

– Suzanne Chutchian

As a therapist and mom, Suzanne regularly gives advice to young women with the condition, urging them not to try to "tough it out" alone, but rather get treatment.

"If you have pain and other symptoms of endometriosis, don't downplay your symptoms or think you're not strong enough. Talk with a woman's health specialist," she says. ■

A daughter's journey

When she was just 10 years old, Rachel Chutchian-O'Sullivan got her first period.

Each month, it lasted for 10 to 14 days, which is considered a long cycle. She also had severe menstrual cramps, pain in her lower spine, and stomach problems.

Rachel's pain was often so bad that she was late for school or missed it altogether.

"I remember walking to the restroom at school and stopping in my tracks," Rachel says. "I couldn't move at all."

"Take us seriously when we complain about horrible pain like this."

– Rachel Chutchian-O'Sullivan

Some days she couldn't focus because the pain was so severe. She was also embarrassed.

"It's not something you want to talk about when you're that young—or ever," Rachel says. Even with a doctor's note, Rachel remembers that her school administrators weren't very understanding about her tardiness or absences due to endometriosis.

Diagnosing the pain

In her early teens, Rachel had numerous doctors visits. Though she suspected it was endometriosis, a condition both her mother and grandmother had, providers wanted to rule out other issues.

They performed several tests, including computed tomography (CT) scans, ultrasounds, a colonoscopy, and an endoscopy.

Getting treatment

Initially, Rachel's pediatrician prescribed birth control pills to help control her menstrual cycle and relieve some of the pain.

"It was still painful, but not as bad as it was before," she says.

Later, Rachel's doctor prescribed a birth control injection, medroxyprogesterone, which also helped ease the pain.

Side effects

The injection did have side effects, including weight gain. Rachel got the injections for several years, but eventually stopped because of a more serious side effect: decreased bone density. So, she went back to taking birth control pills.

"Ever since I was diagnosed, I've been afraid that I won't be able to get pregnant some day," she says. "Knowing that it's hereditary, I fear that if I do have children, I'll pass it on to them, too."

Advice for others

Today, Rachel still experiences cramps, but they're much less severe than when she was younger. At age 25, she's pursuing a master's degree in social work, as her mother did around that age.

She offers the same advice to all people, both women with the condition and the men and women supporting them: "Take us seriously when we complain about horrible pain like this."

She also advises women of all ages who have severe cramps or lower abdominal pain to talk with a women's health specialist as soon as symptoms appear.



Suzanne Chutchian, right, and her daughter Rachel Chutchian-O'Sullivan have both suffered from endometriosis.

"Make it known if you are having issues," Rachel says. "I didn't talk about it when I was younger, but now I do. There's more awareness now." ■

Find Out More

- ▶ **MedlinePlus: Endometriosis**
<https://medlineplus.gov/endometriosis.html>
- ▶ **Eunice Kennedy Shriver National Institute of Child Health and Human Development: Endometriosis**
<https://www.nichd.nih.gov/health/topics/endometriosis>
- ▶ **NIH News in Health: Period Problems**
<https://newsinhealth.nih.gov/2019/08/period-problems>
- ▶ **ClinicalTrials.gov: Endometriosis**
<https://clinicaltrials.gov/ct2/results?cond=endometriosis>

Finding self-confidence with vitiligo



Tonja Johnson created a support group to help others with the common, but difficult, skin condition

Tonja Johnson was diagnosed with vitiligo at age 43 and has lost about 80% of her color.

Tonja Johnson was 41 years old when she noticed a small, white spot on her leg. It wasn't long before she started seeing more spots on both her legs and arms.

As the spots grew in size and number, Tonja decided to see a dermatologist, a health care provider who focuses on skin and hair conditions.

Two years later, at age 43, Tonja was diagnosed with vitiligo. The condition causes some or all of the skin to lose color, leaving white patches.

"I was devastated," Tonja says. "I wondered, 'Why me? Why this disease?'"

Covering up

She tried to cover up her light patches with long-sleeved shirts, but soon her hands started losing pigmentation.

As a dark-skinned African American woman, Tonja says she was teased as a kid for having such dark skin. Then, as an adult, to start losing that very color she had worked so hard to love sent her into a deep depression.

"My vitiligo is aggressive," Tonja says. "I've lost about 80 percent of my skin color in a very short amount of time."

"The very thing that brought so much pain to my life also has brought passion. I no longer feel sorry for myself."

- Tonja Johnson

Her doctor prescribed topical creams to help bring back the color, but Tonja found it hard to keep that up. She lives in Florida where it's hot and humid, so she had to reapply the creams often.

Embracing vitiligo

She considered phototherapy (light therapy) too but it was expensive, costing her several hundred dollars a month. That's when things changed for Tonja.

"About three years ago, I stopped trying to cover it and just embraced it," she says.

Tonja suggests that anyone with vitiligo do their research to understand what it is, how it affects people, and what the treatment options are.

She was surprised by how little information was provided by her doctors and by the lack of support groups, not only in her part of the country but nationwide.

Tonja turned to her family, her friends, and her church for help and support. She also founded her own support group, called Beautifully Unblemished.

"I'm an extrovert, but I became very withdrawn when I started to lose color," she says. "I got a lot of stares and pointing because my nose was white but the rest of my face was black."

Spreading the word

Tonja encourages others to be vocal and spread awareness about this common condition. For instance, she wants people to know it's not just a cosmetic disease—it has real physical and emotional effects.

"It's not contagious—it's an autoimmune disease," Tonja adds.

In addition to spreading awareness and offering support, Tonja also participates in research studies that look at the emotional side of the disease. She hopes this can help others.

Tonja says that her advocacy work and talking about the condition have actually helped her become comfortable in her own skin again.

"The very thing that brought so much pain to my life has also brought passion," she says. "I no longer feel sorry for myself." ■

Is it vitiligo or something else?

Learn the difference between vitiligo and conditions like scleroderma or Addison disease

Vitiligo is not the only disease that causes skin to change color. Here are a few conditions and what they look like, including vitiligo.



Vitiligo is an autoimmune disease that causes your skin to lose color, often resulting in white patches on your skin that cover both sides of your body.



Pityriasis alba is a common skin disorder that causes areas of light-colored skin. The spots often start as slightly red, scaly patches on the face, upper arms, neck, and upper middle section of the body.



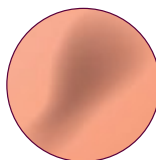
Tinea versicolor is caused by a fungal infection and leads to patches of discolored skin with a fine, dry, scaly surface.



Hansen's disease (leprosy) is a bacterial infection that can also cause discolored patches of skin. Usually these patches are flat, look faded, and may even feel numb.



Scleroderma is a rare but serious autoimmune disease that causes parts of the skin to harden. Often those areas can appear shiny. It can also create problems with connective tissue beneath the skin, harming blood vessels and internal organs.



Addison disease happens when the body doesn't produce enough cortisol, a hormone that regulates stress. One symptom of the disease is darkening of the skin in places like scars, skin folds, elbows, knees, and knuckles.

Talk to your health care provider if you think you might have vitiligo or another skin condition. ■

SOURCE: MedlinePlus

Vitiligo: What you need to know

Causes, treatment options, and finding support



Vitiligo is a skin condition affecting roughly one in 100 people in the U.S. It causes areas of skin to lose pigmentation (color), resulting in light or white patches.

The condition is an autoimmune disease, causing the body's immune system to attack and destroy its own tissue cells. In this case, it attacks skin cells that give the skin pigment.

Where it happens

Vitiligo can happen more frequently on your face and hands, and at pressure points such as your knees and elbows. Vitiligo is a progressive disease that may evolve either slowly or rapidly. While some people develop only a few patches of white skin, others can get patches that cover most of their body.

Two types of vitiligo

- **Vitiligo** is when white patches appear symmetrically on both sides of the body. It is the most common type, accounting for up to 90% of cases.
- **Segmental vitiligo** is much less common. It happens when white patches appear in a localized area on only one side of the body. While it is less responsive to topical treatments like creams, segmental vitiligo generally develops for a short period of time only and then stops, so it is a good candidate for surgical treatments like skin grafts.

Who it affects

Vitiligo is more noticeable in those with darker skin tones, but it can affect people of any race or age.

Treatment options

Researchers supported by the National Institutes of Health are close to getting approval for a new, targeted vitiligo treatment using Janus kinase (JAK) inhibitors. For now, other treatment options include the following:

- **Phototherapy**, also called light therapy, carefully exposes your skin to limited amounts of ultraviolet light. It is recommended for large areas affected by vitiligo.
- **Laser treatment** is recommended for smaller areas of vitiligo, especially those on the face.
- **Steroid creams and ointments** applied to the skin help suppress the immune system.
- **Surgical techniques** include skin grafting, which places pigmented skin from a donor on areas of vitiligo.
- **Depigmentation, or bleaching**, is a permanent change and is used mostly when the full body is affected.

Not every treatment is right for everyone, and many have side effects. But work to discover more broadly effective, longer lasting treatments offers hope.

Finding help

For those with vitiligo, the emotional impact of the condition can be as intense as the physical effects. The condition is not contagious or life-threatening, but it can cause low self-esteem.

People with vitiligo can become depressed and withdraw from friends and family. It's important for those with the condition to find support from friends or loved ones, counselors, or support groups. ■

SOURCES: MedlinePlus; National Institute of Arthritis and Musculoskeletal and Skin Diseases; American Academy of Dermatology; Genetics Home Reference

FAST FACTS

About **15% to 25%** of people with vitiligo are also affected by **at least one other autoimmune disorder**.

SOURCE: Genetics Home Reference

Treating vitiligo: Studies look for long-term options

JAK inhibitors and immunosuppressants hold promise



Researcher John E. Harris, M.D., Ph.D., has discovered breakthrough treatment options for vitiligo.

“MANY PEOPLE BELIEVE THAT VITILIGO IS A RARE DISEASE,”

says National Institutes of Health-supported researcher John E. Harris, M.D., Ph.D. “It’s not. It’s one of the most common diseases, affecting one in 100 people worldwide.”

“We want to keep the research going and get to a point where people can decide whether they want to embrace their spots—or not.”

– Caroline Le Poole, Ph.D.

Decades of research by professors Caroline Le Poole, Ph.D., and Dr. Harris have helped to reveal the central underpinnings of this condition.

Fifteen to 20 years ago, the autoimmune nature of vitiligo began to surface. Since then researchers have been developing an increasingly more accurate map about underlying mechanisms. This is now allowing the development of several treatments to disrupt vitiligo.

Researching more treatments

While there’s no cure for vitiligo yet, there are very good treatments available today. That said, Dr. Harris, Dr. Le Poole, and other researchers are working hard to find ones that are even more effective.

“There are now four clinical trials testing Janus kinase (JAK) inhibitors,” Dr. Harris notes. There were none just a few years ago.

“We’re very close to having a U.S. Food and Drug Administration-approved treatment,” he adds.

JAK inhibitors work to prevent the signaling of specific proteins that cause vitiligo. This kind of targeted treatment is promising, but it’s still not a cure. When people with vitiligo stop the drug, the disease comes right back, in the same places it was before.

Dr. Harris and Dr. Le Poole have recently learned that they may be able to stop memory cells from allowing the disease to come back.

“Now that would be a total game changer,” Dr. Harris says. “It would mean we could treat people for a short time and get years’ worth of benefits.”

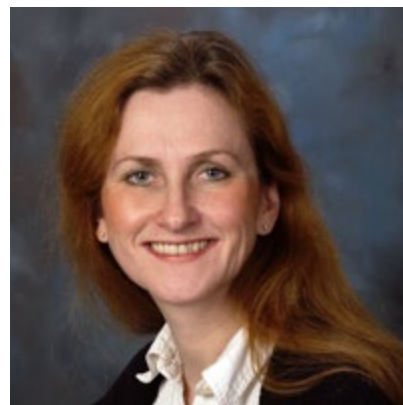
Dr. Le Poole says their research teams are also looking to start a clinical trial for an immunosuppressant, known as a heat shock protein, that could help reverse vitiligo.

“The treatment is meant to nip the development of the disease in the bud,” says Dr. Le Poole. “There are new options on the horizon.”

Providing options

As research continues to advance, many people with vitiligo are also embracing, rather than treating, their patches. Celebrities, such as model Winnie Harlow, and other advocates have helped inspire confidence and spread awareness.

“People are seeing the beauty in their spots,” Dr. Le Poole says. “Patients are more expressive than ever before. They’re presenting their disease in an open way and are being empowered to accept their skin exactly how it is.”



Vitiligo researcher Caroline Le Poole, Ph.D., wants to give patients more options.

Dr. Le Poole and Dr. Harris still want to give people with vitiligo as many treatment options as possible, so they can choose what’s best for them.

“We want to keep the research going and get to a point where people can decide whether they want to embrace their spots—or not,” Dr. Le Poole adds. ■

Find Out More

- ▶ **MedlinePlus: Vitiligo**
<https://medlineplus.gov/vitiligo.html>
- ▶ **National Institute of Arthritis and Musculoskeletal and Skin Diseases**
<https://www.niams.nih.gov/health-topics/vitiligo>
- ▶ **ClinicalTrials.gov: Vitiligo**
<https://clinicaltrials.gov/ct2/results?cond=vitiligo>

from
the

lab

LATEST
RESEARCH
UPDATES
FROM NIH

New NIH study may help predict those at risk for severe MS

Dark-rimmed spots in the brain may signal a severe form of the disease

RESEARCHERS MAY SOON be able to look at images of our brains to help diagnose multiple sclerosis (MS) earlier.

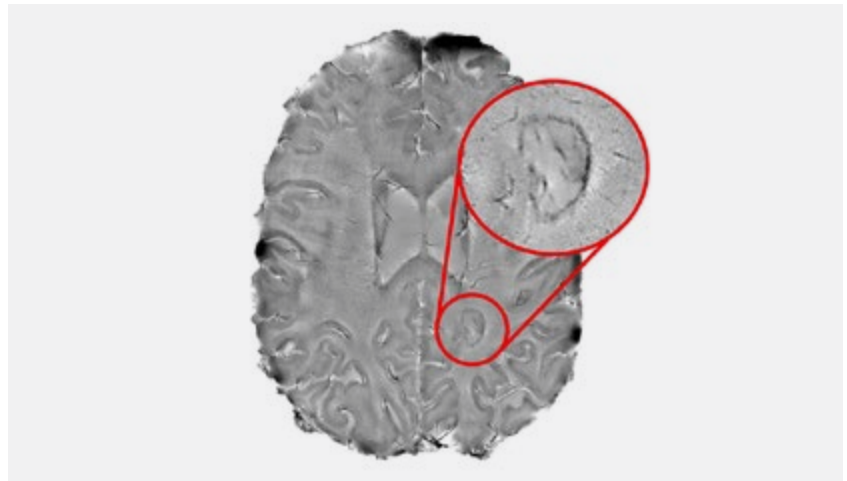
MS affects more than 2 million people worldwide. The disease happens when the body's immune system attacks the protective coating around nerve cells in the brain and spinal cord.

In a recent study, National Institutes of Health (NIH) researchers followed 192 people with MS for seven years. They found that more than half of the patients had one or more dark-rimmed spots inside their brains. These spots may be markers for a more serious form of the disease.

Some early symptoms of MS include problems with seeing, balance, and muscle strength. But more aggressive forms have symptoms such as paralysis and serious problems with thinking and memory.

That's why researchers at NIH and researchers around the country are working hard to help detect the disease early.

The new study was conducted with patients at the NIH Clinical Center, the largest research hospital in the country. There, NIH researchers led by Daniel S. Reich, M.D., Ph.D., used a



Dark-rimmed spots on the brain, like this one, may suggest a more severe form of multiple sclerosis and help with early detection.

powerful magnetic resonance imaging (MRI) scanner to take pictures of the brains of the patients with MS.

His team then used a 3D printer to compare the dark-rimmed spots in the scans with similar spots seen in brain tissue samples. They found that patients with four or more of these spots were more likely to have the aggressive form of MS than those without them.

“Our results point the way toward using specialized brain scans to

predict who is at risk of developing progressive MS,” Dr. Reich says.

His team previously published instructions for clinics on reprogramming their lower-powered MRI scanners to better detect these spots. The team hopes that researchers around the world will use these instructions to develop better diagnostic and treatment strategies for people with MS. ■

SOURCES: National Institute of Neurological Disorders and Stroke; NIH Research Matters



Pregnant women may want to avoid air pollution in days before delivery

NIH study suggests potential link to increased NICU admissions

AIR POLLUTION can cause problems for many people—but it may be especially harmful in pregnant women.

National Institutes of Health researchers found that pregnant women who are exposed to certain air pollutants may be more likely to have babies admitted to the neonatal intensive care unit (NICU).

Polluted air may cause inflammation in the mother's body. This could harm the placenta, the tissue that connects the mother and the fetus.

The research included data from 220,000 women who gave birth between 2002 and 2008. About 27,000 of those women had babies admitted to the NICU.

Researchers looked at air quality data in the area where each birth occurred and when the exposure to polluted air happened, for example, the week before delivery, the day before delivery, and the day of delivery.

The chances of NICU admission increased significantly if mothers were exposed to traffic-related air pollution on the day before and the day of delivery.

More research needs to be done to confirm the study's results. For now, pregnant women may want to limit their time outdoors during air quality advisories near the end of pregnancy. ■

SOURCES: Eunice Kennedy Shriver National Institute of Child Health and Human Development; NIH Research Matters

Taking a daily aspirin no longer recommended for many older adults

New guidelines recommend it just for heart attack and stroke survivors

MANY PEOPLE ARE TAKING a daily low-dose aspirin even though they don't need to.

New National Institutes of Health-supported research suggests that nearly 29 million people may be taking aspirin unnecessarily to help prevent a heart attack or stroke. Ten million of those people are 70 and older.

In the past, experts recommended that people with heart problems take one low-dose aspirin a day. Aspirin helps thin the blood to prevent blood clots, which can lead to a stroke or heart attack.

But new guidelines recommend that only for heart attack or stroke survivors.

Researchers also found that people were taking aspirin even though their doctor hadn't recommended it. This can be especially dangerous in people who are at higher risk of internal bleeding, like those with stomach ulcers.

Make sure to check with your health care provider before starting or stopping any medicine, even one that is available without a prescription. Your pharmacist can also tell you which medications may not work well together. ■

SOURCES: National Heart, Lung, and Blood Institute; NIH Research Matters



NIH on the web

Online resources for safe infant sleep

➔ **DID YOU KNOW** that blankets shouldn't be used in cribs for babies under 1 year old?

This is one of many tips that can help reduce the risk of Sudden Infant Death Syndrome (SIDS). The condition happens when a baby younger than 1 year old dies without a clear cause.



To help raise awareness and educate caregivers about SIDS, the Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD) has developed a number of online resources. All are available on NICHD's Safe to Sleep® website and

include up-to-date information reviewed by NIH researchers.

You can see a diagram of what a safe sleeping environment should look like or watch videos tailored to the type of caregiver, like breastfeeding moms or grandparents.

Find it all in one place!

magazine.medlineplus.gov/otw



Vision loss meets virtual reality

➔ **EVER WONDERED** what vision loss is like?

The National Eye Institute has created a new app that gives you a taste through virtual reality. The app, See What I See, lets users experience effects of age-related macular degeneration or cataracts on their phone or tablet.

Those include blurriness and spots in your vision.

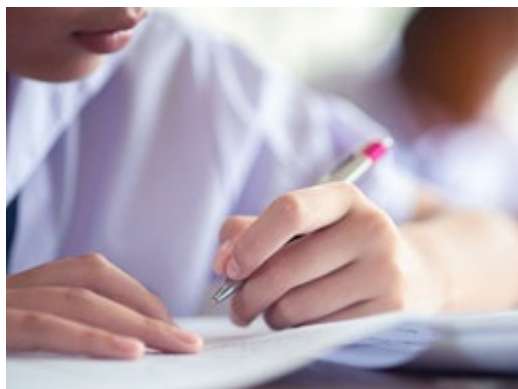
Users can choose from scenes, like a city street at night, and then see how it looks to someone with vision problems. The app is available for free on the Apple Store and Google Play.

Students speak up about mental health

➔ **A NATIONAL ESSAY CHALLENGE** encouraged students to address the challenges of stigma, fear, and improving mental health education and treatment, particularly in vulnerable populations.

The National Institute of Mental Health and the National Institute on Minority Health and Health Disparities (NIMHD), in collaboration with the Calvin J. Li Memorial Foundation, sponsored the contest and selected winners from more than 160 entries from high school students across the U.S. and Puerto Rico.

Take a look at the 12 winning and honorable mention essays on the NIMHD website.



NIH is here to help

The National Institutes of Health (NIH)—the nation’s medical research agency—includes 27 Institutes and Centers and is a part of the U.S. Department of Health and Human Services. It is the primary federal agency for conducting and supporting basic, clinical, and translational medical research, and it investigates the causes, treatments, and cures for both common and rare diseases. For more information about NIH and its programs, visit www.nih.gov.

Institutes

National Library of Medicine (NLM)

www.nlm.nih.gov
888-FIND-NLM 888-346-3656

National Cancer Institute (NCI)

www.cancer.gov
800-4-CANCER 800-422-6237

National Eye Institute (NEI)

www.nei.nih.gov
301-496-5248

National Heart, Lung, and Blood Institute (NHLBI)

www.nhlbi.nih.gov
301-592-8573

National Human Genome Research Institute (NHGRI)

www.genome.gov
301-402-0911

National Institute on Aging (NIA)

www.nia.nih.gov
Aging information 800-222-2225
Alzheimer’s information 800-438-4380

National Institute on Alcohol Abuse and Alcoholism (NIAAA)

www.niaaa.nih.gov
301-443-3860

National Institute of Allergy and Infectious Diseases (NIAID)

www.niaid.nih.gov
301-496-5717

National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS)

www.niams.nih.gov
877-22-NIAMS 877-226-4267

National Institute of Biomedical Imaging and Bioengineering (NIBIB)

www.nibib.nih.gov
301-451-6772

Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD)

www.nichd.nih.gov
800-370-2943

National Institute on Deafness and Other Communication Disorders (NIDCD)

www.nidcd.nih.gov
800-241-1044 (voice)
800-241-1055 (TTY)

National Institute of Dental and Craniofacial Research (NIDCR)

www.nidcr.nih.gov
301-480-4098

National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK)

www.niddk.nih.gov
NIDDK Health Information Center
1-800-860-8747

National Institute on Drug Abuse (NIDA)

www.nida.nih.gov
301-443-1124

National Institute of Environmental Health Sciences (NIEHS)

www.niehs.nih.gov
919-541-3345

National Institute of General Medical Sciences (NIGMS)

www.nigms.nih.gov
301-496-7301

National Institute of Mental Health (NIMH)

www.nimh.nih.gov
866-615-6464

National Institute on Minority Health and Health Disparities (NIMHD)

www.nimhd.nih.gov
301-402-1366

National Institute of Neurological Disorders and Stroke (NINDS)

www.ninds.nih.gov
800-352-9424

National Institute of Nursing Research (NINR)

www.ninr.nih.gov
301-496-0207

Centers & Offices

Fogarty International Center (FIC)

www.fic.nih.gov
301-496-2075

National Center for Complementary and Integrative Health (NCCIH)

www.nccih.nih.gov
888-644-6226

National Center for Advancing Translational Sciences (NCATS)

www.ncats.nih.gov
301-435-0888

NIH Clinical Center (CC)

clinicalcenter.nih.gov
301-496-2563

Office of AIDS Research (OAR)

www.oar.nih.gov
301-496-0357

Office of Behavioral and Social Sciences Research (OBSSR)

www.obssr.od.nih.gov
301-402-1146

Office of Communications & Public Liaison (OCPL)

www.nih.gov/institutes-nih/nih-office-director/office-communications-public-liaison
301-496-5787

Office of Rare Diseases Research (ORDR)

www.rarediseases.info.nih.gov
Genetic and Rare Disease
Information Center
888-205-2311

Office of Research on Women’s Health (ORWH)

orwh.od.nih.gov
301-402-1770

*It's time
to rewrite
the story of
sickle cell.*



**CURE
SICKLE
CELL.**

Add your voice at
CURESICKLE.ORG



National Heart, Lung,
and Blood Institute