

WINTER 2019

# NIH MedlinePlus

MAGAZINE

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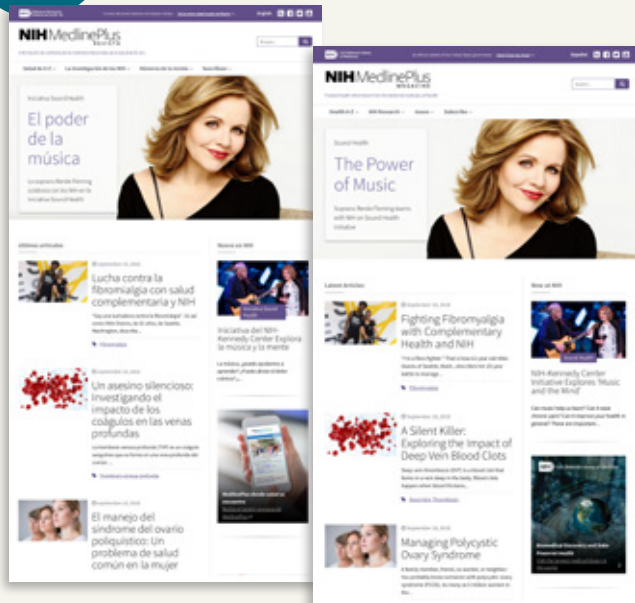
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# SCLERODERMA

new  
website

coming soon!



## New Year, New Opportunities for NIH MedlinePlus Magazine and You!

**W**E'RE WORKING HARD TO BRING YOU MORE of the NIH MedlinePlus magazine content you love, in fresh and exciting new ways.

We're adding additional features and a fresh design to our NIH MedlinePlus magazine website, [magazine.medlineplus.gov](http://magazine.medlineplus.gov).

You will soon be able to search the magazine site by MedlinePlus health topics and access additional articles on topics you care about. That includes more personal stories from patients and everyday people living with conditions, and articles from celebrities and advocates sharing their own health experiences.

You will also be able to easily navigate to MedlinePlus on every page of the site for deeper dives on health topics, diseases and conditions, research updates, and treatment information.

The site will feature easy-to-navigate menus, scrolling social media share buttons, and a mobile-friendly design that works seamlessly on your phone, tablet, or desktop.

We are also working to make the website fully bilingual. Soon, you will be able to read articles in both English and Spanish.

Our goal is to provide more readers with access to trusted health information from NIH, the National Library of Medicine, and MedlinePlus.

Stay tuned for more updates as we roll out these new and improved website features. We look forward to continuing this journey with you! ■

**NIH MedlinePlus**  
MAGAZINE

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24 Superior Drive, Suite 103  
Natick, MA 01760 | (508) 907-7000

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## 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 the world's largest medical library.

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Thanks for reading!

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Comedian Bob Saget (pictured above with “Fuller House” co-stars Dave Coulier and John Stamos) shares his personal experience with scleroderma advocacy.

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Helping your child through winter ear infections

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# health

NEWS,  
NOTES,  
& TIPS  
FROM NIH

## Help Your Child Through Winter Ear Infections

**HEALTH TIPS** The onset of winter often means the worst part of cold and flu season, and with that, the dreaded ear infection.

An ear infection is a bacterial or viral infection that affects the middle ear. The infection is often painful because it creates pressure in the small space between the eardrum and the back of the throat (the eustachian tube).

Unfortunately, ear infections affect young children more than others. Eustachian tubes are smaller and more level in infants and young children, making it more difficult for fluid to drain out of the ear. When tubes become blocked with mucus, it creates pressure on the eardrum.

Because a child's immune system is not as developed as an adult's, it may be harder to fight a sore throat, cold, or respiratory infection that can trap fluid behind the eardrum.

Older children and adults can also get ear infections, but they are less common because their eustachian tubes are larger and slanted, so they drain fluid better.

### What to look for in your child

If your child is not old enough to communicate pain, here are a few signs to look for:

- Tugging or pulling at the ear(s)
- Irritability and crying more than usual
- Trouble sleeping
- Fever (especially in infants and young children)
- Fluid draining from the ear
- Loss of balance
- Difficulty hearing or responding to quiet sounds

Your child's health care provider can check for an ear infection by using an otoscope. The lighted tool shows redness in the ear, fluid, or less likely, a rupture in the eardrum.



They will typically ask if your child has had a runny nose, cough, or fever, since the virus or bacteria that cause the cold can spread to the middle ear.

### What to do

Some ear infections clear up on their own. However, if an antibiotic is prescribed, it is important that your child takes the medicine for the full time that your doctor advises. Typical treatment lasts seven to 10 days.

If your child has repeated ear infections, your health care provider may suggest draining the ear with small ventilation tubes. The tubes improve air flow and prevent fluid backup in the middle ear so that the child can hear better.

### Reducing the risk

Here are ideas to help your child get through winter without an ear infection:

- Stay up to date with all vaccinations, including the flu shot.
- Frequently wash hands.
- Avoid cigarette smoke, as studies show that babies who

are around smokers have more ear infections.

- Limit your child's exposure to sick playmates.
- Avoid baby bottles in the crib, as fluid can accidentally back up in the middle ear.

### What's next?

Researchers sponsored by the National Institute on Deafness and Other Communication Disorders are exploring vaccines against some of the most common bacteria and viruses that cause middle ear infections.

They are also learning more about colonies of antibiotic-resistant bacteria, called biofilms, that are in the middle ears of most children who have multiple ear infections. Restricting the nutrients that grow the biofilms may fight the bacteria. ■

**SOURCES:** MedlinePlus; NIH News in Health; National Institute on Deafness and Other Communication Disorders

### DID YOU KNOW?

**5 out of 6**

**Five out of six children** will have at least one ear infection by their third birthday.



# HPV Vaccine Approved for Ages 27 to 45

**BY THE NUMBERS** January is Cervical Health Awareness month, and there is good news on the protection front for one cause of cervical cancer: the human papillomavirus, or HPV.

Last fall the Food and Drug Administration approved the Gardasil 9 vaccine for men and women ages 27 to 45. Gardasil is also known as the HPV 9-valent vaccine, recombinant. It protects against nine types of HPV.

Both men and women can get HPV and pass it on without knowing it. Additionally, there is low-risk HPV and high-risk HPV. Low-risk HPV can cause genital warts. High-risk HPV can cause cancers of the cervix, vagina, penis, anus, and back of the throat.

HPV vaccination can prevent more than 90 percent of cervical cancers from ever developing by preventing the infections that cause those cancers.


There are more than **200 types of HPV viruses**. More than 40 of them can easily spread through direct sexual contact.



Every year about **14 million Americans** become infected with HPV.

It can take between **10–30 years** from the time of an initial HPV infection until a tumor forms.



**33.7k+** 

HPV causes more than **33,700 cases of cancer** every year in the U.S.

**SOURCES:** MedlinePlus; Centers for Disease Control and Prevention; Food and Drug Administration

# NIH Collaborates on National Limb Loss Registry

*Project with Department of Defense aims to improve quality of care*

**NIH RESEARCH** Limb loss is an important medical issue, especially for U.S. military service members. Now the first national database on limb loss is planning to open in 2020—with help from NIH.

The Limb Loss and Preservation Registry will help the medical and military communities better understand amputee challenges and needs. Ultimately it aims to improve the care of those who have experienced limb loss.

NIH is partnering with the Department of Defense (DoD) on the project, which will be led by the Mayo Clinic, an academic medical center.

“The Limb Loss and Preservation Registry addresses a significant public health knowledge gap,” says Alison Cernich, Ph.D. She is director of the National Center for Medical Rehabilitation Research within NIH’s *Eunice Kennedy Shriver* National Institute of Child Health and Human Development (NICHD).

NICHD is leading the limb loss registry effort within NIH.

## Understanding limb loss

Limb loss can happen for many reasons. Those include birth defects, surgical procedures, or traumatic injury, such as in military combat.

Researchers studying diseases and conditions that can contribute to limb loss, such as vascular disease and diabetes, will have access to the registry, says Dr. Cernich. Researchers will be able to sort the data by age, gender, and type of limb loss.



*The registry will help improve treatment and rehabilitation for people who have lost limbs.*

### Information from the database will help:

- Prevent limb loss
- Improve amputation surgeries
- Refine rehabilitation approach
- Improve devices for people with limb loss

## Partnering to understand limb loss

NIH is partnering with the DoD on developing the registry in an effort to improve the quality of care for active military members and veterans, along with other limb loss patients.

“The joint effort between federal agencies allows us to collect data that will inform research and improve the lives of all citizens coping with limb loss,” Dr. Cernich says. ■

**To find out more about limb loss, visit MedlinePlus.**

**SOURCE:** NIH News Releases

# New MedlinePlus Topic: Acute Flaccid Myelitis

**NEW ON MEDLINEPLUS.GOV** Acute flaccid myelitis has made headlines recently, but what exactly is this strange disease that affects children?

Using MedlinePlus' new health topic page, we break down what you need to know about this rare but serious illness that affects the nervous system.

It's a neurologic disease that affects the spinal cord. Neurologic diseases involve the brain, spinal cord, and nerves, which make up the nervous system.

- Symptoms include sudden arm or leg weakness and loss of muscle tone and reflexes.
- It often happens after a mild viral respiratory illness, like a cold, or a fever.
- Anyone can get it, but recent cases have been in children.

The condition is not new. However, there have been more cases reported since 2014.

Enterovirus and West Nile virus can all cause acute flaccid myelitis.

If you suspect you or a loved one has acute flaccid myelitis, see your health care provider immediately. They may perform neurological exams and other tests if it is suspected. ■

**SOURCES:** MedlinePlus; Centers for Disease Control and Prevention



*The polio-like disease can cause symptoms such as sudden arm or leg weakness.*



## Did You Know?

### Nobel Prize Winners at NIH

Out of just 12 Nobel Prize winners in 2018, three were NIH-supported researchers. And they're not the first.

- NIH grantees have won Nobel Prizes dating back to 1939.
- 156 NIH-supported researchers have been sole or shared recipients of 92 Nobel Prizes.
- Want to see a Nobel Prize in person? There's one on permanent display at the National Library of Medicine's History of Medicine Division. The prize was won by Marshall Nirenberg, who helped us understand genetic code.

**SOURCES:** NIH Almanac; NIH News Releases

# Zoey's Story:

## One Family's Experience with a Rare Disease

*Undiagnosed urea cycle disorder takes life of college student*

In 2015, college student Zoey Zalusky was 19 years old with an exciting future ahead of her. She had a supportive, loving family, and a close circle of friends. She was studying to be a nurse and getting good grades at the University of Arizona at Tucson.

"She was our social butterfly," says Renee Zalusky, Zoey's mom. "She was always everywhere and wanting to do everything. She had a lot of energy." Zoey was the middle of three children.

But in December of that year, everything changed. Zoey suddenly passed away from an undiagnosed, rare genetic condition: ornithine transcarbamylase deficiency (OTC), which is part of a larger group of urea cycle disorders.

The disorder can go unnoticed for years, as it did in Zoey's case. If not diagnosed, it can lead to a buildup of toxic levels of ammonia in the body, which can cause brain damage and other serious complications. Ammonia builds up because people with the disorder cannot dispose of excess nitrogen—which comes from the protein we eat and the protein in our muscles—through their urine.

### Zoey's childhood

Renee says looking back there were some symptoms, but they didn't seem out of the ordinary at the time. In general, Zoey was a healthy, happy child.

"As far as health goes, I always described her as energetic and full of life," Renee says. "Whenever we'd go to her annual check up, there were never any problems." Zoey played sports growing up and swam on the varsity swim team in high school.

But Zoey did have an aversion to protein from the time she was little and always preferred carbohydrates and salads. With urea cycle disorder, people often avoid protein because it can

make them feel sick. In Zoey's case, it just seemed like something a lot of kids do: picky eating.

"Zoey never ate a burger in her life. She never ate a steak," Renee says. "It's easy to see now that she had a definite aversion to protein because it probably made her feel sick."

As she got closer to adolescence, Zoey started to have uncontrolled episodes of vomiting. It only happened once in a while, with no warning, sometimes during high stress periods, Renee says.

"She would just out of the blue suddenly vomit. Sometimes she knew it was going to happen and sometimes she wouldn't," Renee says.

Her health care providers at the time thought it might be food allergies or sensitivities. They suggested eliminating certain food groups and then re-introducing them slowly, to see if they could determine what was causing her nausea and vomiting.

Overall, they did not seem concerned about the situation and Zoey never really took the diet modifications seriously.

### 2015

Zoey's disease went unnoticed until the winter of 2015.

It was the first semester of her sophomore year, and Zoey didn't feel well. She called her mom (who lives in Texas) saying she fell asleep at 4 p.m. the day before and had slept until 9:00 a.m. the next morning. Her words were jumbled and she wasn't making sense.

"She was so upset because she was trying to tell me she had missed an assignment the day before," Renee says. "At first I thought she might have been drinking."

Hours later, Zoey wound up in the emergency room at the college's hospital. Her doctors considered alcohol poisoning, but after doing a variety of tests, including drug and alcohol screening, they weren't able to diagnose anything in particular.







Looking back, Zoey was having a reaction to the ammonia buildup in her body due to urea cycle disorder.

“They came to the conclusion that she was mildly dehydrated,” Renee says. “The only thing that really raised some red flags were that her liver enzymes were slightly elevated.”

The doctors gave her fluids, which seemed to help. Zoey was told to come back a few days later for a checkup and to follow up with her family doctor.

Zoey had a few repeat blood tests, which showed her liver enzymes were still high. But overall, she seemed back to normal and tried to catch up on schoolwork, Renee says.

But a few weeks later, Zoey collapsed. Her urea cycle disorder caused her to have irreversible brain damage due to the high levels of ammonia in her body, which led to a fatal coma.

## Discovering urea cycle disorder

After Zoey passed away, her parents made the decision to donate her organs. On Christmas Day 2015, three people received the gift of lifesaving organs. This is how they eventually discovered what led to Zoey’s coma.

The Arizona Donor Network contacted them, letting them know that the man who received Zoey’s liver had died shortly after due to complications from urea cycle disorder.

Zoey’s whole family (her parents and two siblings) were all tested at Baylor College of Medicine in Houston after hearing this. They discovered that Zoey’s father, John, also has OTC deficiency. The disease had developed spontaneously in utero, or before he was born.

## Spreading the word

Zoey’s family still works closely with doctors at Baylor, who help John manage his OTC deficiency. Baylor also helped connect them with the National Urea Cycle Disorders Foundation (NUCDF), a patient advocacy group. They’re

“Zoey never ate a burger in her life. She never ate a steak. It’s easy to see now that she had a definite aversion to protein because it probably made her feel sick.”

- Renee Zalusky

committed to telling Zoey’s story to help others and share her memory. A special part of this was a video discussing Zoey’s life and her OTC story.

Renee is also spreading the word about blood ammonia tests, which can help diagnose urea cycle disorders. Zoey’s ammonia level was not tested during either of her hospital stays.

“It’s not an expensive test to do, but it’s not an easy one either,” Renee says.

For an ammonia test, the blood sample must be run immediately, because after 30 minutes there is a risk of a false positive.

The NUCDF has created a website that seeks to address this lack of awareness among medical professionals about the hidden dangers of ammonia poisoning.

“It was so devastating for us that nobody had ever really heard of this disorder, and it became really important to tell people Zoey’s story,” Renee says. “We’ve been trying to do whatever we can to support efforts to educate the public, as well as emergency room personnel. If even one family can be saved from what happened to us, it will be worth it.” ■

# What is it Like to Research a Rare Disease?

## UREA CYCLE DISORDER PRINCIPAL INVESTIGATOR SHARES HER EXPERIENCE

*Andrea L. Gropman, M.D., studies rare diseases with support from NIH and the Rare Diseases Clinical Research Network. She talked about what it's like to research a rare disease as a principal investigator on the Urea Cycle Rare Diseases Consortium. She has been studying the disorder for 14 years.*

### What is a urea cycle disorder?

A urea cycle disorder results from an inability to safely dispose of nitrogen in the body. When our bodies break down protein, we create nitrogen that is used for many chemical reactions in the body for growth, and excess nitrogen is made into a nontoxic chemical called urea that we get rid of through our urine.

“Patients actually provide a lot of valuable suggestions to us as researchers because they are living with the disease.”

- Andrea L. Gropman, M.D.

But when that process goes wrong, in people with urea cycle disorders, the excess nitrogen is not flushed out through our urine. Instead, the nitrogen builds up as ammonia in our bodies and causes damage, particularly in the brain.

### How do people get the disorder?

Urea cycle disorders are genetic conditions. They can arise from inheriting a mutated gene from the

parents or it may start as a new condition in the patient.

### How many patients have a urea cycle disorder?

We think that it's about 1 in 30,000. We don't know the exact number of patients out there because many individuals remain undiagnosed or have very mild symptoms, especially adults.

### What kind of research does the Urea Cycle Rare Diseases Consortium work on?

The goals of our consortium are to better understand what causes urea cycle disorders, to study the natural history of these disorders, and develop new treatments. Already, our consortium has improved the care of adults and children with urea cycle disorders. We are now looking at related issues, like liver conditions that may develop later in life.

We're currently working on expanding newborn screening to all urea cycle types (there is screening available for some of the types of urea cycle disorders now) so they can be identified before brain injury has set in.

### Can you tell us some specifics?

My project in particular uses magnetic resonance imaging (MRI) to look at the long-term effects of hyperammonemia on the brain. People's attention and their working memory are impacted by urea cycle disorder and there are changes in brain structure and networks that we can study using MRI.

### What's something we may not know about urea cycle disorder research?

Many of our researchers have been working for decades on urea cycle disorders. And we also have junior investigators who are training under



**Andrea L. Gropman, M.D. researches urea cycle disorders with funding from NIH. She works at Children's National Health System in the Washington, D.C. area.**

more senior researchers to carry on this research. That's one of the basic requirements of the network, that there be a substantial commitment to educating the next generation of physicians and researchers in rare disease.

### What advice do you have for patients of rare diseases?

For a patient or the family of a patient, it's often helpful to align yourself with advocacy groups for that disorder. Those groups are partners with us in our research design and study and are very supportive of families.

Patients actually provide a lot of valuable suggestions to us as researchers because they are living with the disease. We always want to hear from patients.

And for patients who may think they have symptoms of a rare disease, they should discuss that with their physician and possibly bring information with them, such as family history, unusual dietary preferences, or unusual blood work results. ■

# Supporting Rare Disease Research at NIH

Rare Diseases Clinical Research Network brings the spotlight to over 7,000 conditions

*Tiina K. Urv, Ph.D., leads the Rare Diseases Clinical Research Network, part of NIH's Office of Rare Diseases Research. She discussed the network, which includes 22 groups across the country and conducts research on approximately 200 rare diseases, with NIH MedlinePlus magazine.*



Tiina Urv, Ph.D., works to support rare disease research throughout the country.

## What makes a rare disease rare?

We define rare diseases as defined in law, through an amendment to the Orphan Drug Act of 1983. That is a condition that affects fewer than 200,000 Americans.

## What is the biggest misconception about rare diseases?

The biggest misconception is that rare diseases are rare. There are over 7,000 rare diseases that impact the lives of over 30 million people.

If you look at it collectively, that's as many people in the U.S. who have diabetes, and as many as those with HIV, cancer, and Alzheimer's combined. Rare diseases are a huge public health problem and we need to start thinking about them as a group as opposed to one at a time.

## Can you tell us about the Rare Diseases Clinical Research Network?

The network came about through legislation that passed in November of 2002, known as the Rare Diseases Act.

Since 2003, the network has funded 31 individual consortia (smaller research groups within the network), touched on over 238 disorders, and over 40,000 people have participated in one way or another in the network over time.

## What areas of research are promising for rare diseases?

Everyone within the network is doing something a little different. For instance, gene editing is really exciting. We have a lot of hope with that.

## How do you recruit people with rare diseases for clinical trials?

It is challenging. That's one of the things that the network works on. How to best go out and find patients. They work closely with the patient advocacy groups.

Each of the individual groups consists of a team that includes patients, advocacy groups, researchers, and clinicians. The individual groups each focus on at least three

rare diseases. These consortia work very closely with NIH and its Rare Diseases Clinical Research Network.

## How do you recommend patients or their loved ones with rare diseases get involved or join a study?

A really good resource we have at NIH is the Genetic and Rare Disease Information Center (GARD). It links you to so many wonderful resources. If you're looking for a family group, you can either email them or call them. We can help them find the care they need as best we can. If somebody's looking for a clinical trial, all clinical trials have to be registered at [clinicaltrials.gov](http://clinicaltrials.gov). And the Rare Diseases Clinical Research Network has a patient registry.

## What message do you have for patients struggling with a rare disease?

You're not alone. There are people who care and there are people out there—researchers, clinicians, patient advocacy groups—who are really working hard to make a difference and find answers. It's a really amazing community. ■

## Find Out More

- ▶ **MedlinePlus: Rare Diseases**  
<https://medlineplus.gov/rarediseases.html>
- ▶ **Rare Diseases Clinical Research Network**  
<https://www.rarediseasesnetwork.org/>
- ▶ **National Urea Cycle Disorders Foundation**  
<http://www.nucdf.org/>

# Don't Give Up Hope

Bob Saget's Unlikely Path to Scleroderma Advocacy

*Television actor and comedian Bob Saget has been making Americans laugh for decades. The “Full House,” “America’s Funniest Home Videos,” and “Fuller House” star has also won praise for his longtime advocacy on behalf of people battling scleroderma, a chronic autoimmune disease. He recently spoke with NIH MedlinePlus magazine about his advocacy and commitment to finding a cure for the disease.*



Bob Saget on the set of “Fuller House,” left, and right with other comedians and entertainers at an event for scleroderma awareness.

“Research is the key that opens the door for the cure. You cannot eradicate the disease or diminish the disease without understanding it.”

— Bob Saget

#### How did you first get involved as an advocate for those with scleroderma?

It’s an unusual story. One day I got a call from someone I did not know asking me to host a comedy fundraiser for a disease I knew very little about. The call was from Sharon Monskey, a wonderful and amazing woman, a former ice skater who had scleroderma and founded the Scleroderma Research Foundation (SRF).

I said yes and hosted the event, which starred Ellen DeGeneres, Rosie O’Donnell, and others. Little did I know that just a few years later, my sister would be diagnosed with the disease.

I have been on the board of directors of the SRF for over a decade now and hosted their events for 25 years.

#### Your sister fought a long battle with scleroderma. Can you share her story with us?

My sister, Gay Saget, was a school teacher near Philadelphia. She was 44 when she was diagnosed with systemic scleroderma. She got treatment, but it was just treating her symptoms with drugs like prednisone and cortisone. She had to move to Los Angeles to live with my parents because she needed so much help. She passed away just two years later.

The good news is that since then we have made some remarkable progress. There are new drugs specifically for scleroderma that are helping people. But we have a long way to go to get to even more effective treatments and eventually a cure.

#### What message do you have for those living with the condition and their loved ones?

I speak with and meet a lot of people with the condition.

My word to them is don’t give up hope because we are making incredible progress.

I also advise them to get educated about the condition and to find a real expert in scleroderma to care for them. That is key. And if they can, try to get help from a center of excellence to get the best treatment.

It is incredibly painful to have a loved one experience a condition like this. It is a very painful disease. My family is still having post-traumatic stress disorder. I don’t know how my parents endured. But I would tell loved ones: Don’t give up hope. Stand by them and get them help. And get them help as early as possible.

#### Why is research like that supported by NIH so important?

There is no improved treatment and there is no cure without the research. Research is the key that opens the door for the cure. You cannot eradicate the disease or diminish the disease without understanding it. We have some of the greatest minds in science working on this. And when you unlock the gate on scleroderma, it will impact a lot of other conditions.

#### What does the future hold for you?

I’m busy! First, I will for the rest of my life remain committed to finding a cure for scleroderma through my work with SRF. We had the most recent “Cool Comedy, Hot Cuisine” fundraiser in December.

Outside of that, I’m happy that Netflix is now airing Season 4 of “Fuller House.” We got nominated for an Emmy. We lost to “Sesame Street.” You can’t get mad at “Sesame Street!” My latest comedy special is called “Zero to Sixty” on Amazon. I’m directing a documentary on the comic Martin Mull. And I’m about to go on tour around the country with my stand-up comedy for a long run. Also, I’m currently in pre-production for a new show on ABC called “Videos After Dark,” which is a more adult, edgy version of the old video show I used to host and write. ■

# Scleroderma: An Overview for You

The name scleroderma means “hard skin”—a simple description of a complex disease that causes thick, hard scar tissue to form either on the outer skin or inside the body around major organs.

In serious cases, this scarring can cause serious damage to the lungs, kidneys, digestive tract, and heart. A variety of medications can help control symptoms and slow further damage, but so far there is no cure. Recent NIH-funded research suggests that stem cell transplants may provide hope for some cases.

## What causes scleroderma?

Scleroderma is considered an autoimmune disorder, which means the body is being attacked by its own immune system. In this case, the process of forming scar tissue—the immune system’s natural response to help heal and protect damaged skin—has gone wrong.

The body overproduces collagen, which causes thick, tight patches of skin that stiffen, squeeze, and ultimately damage portions of the body. What causes this malfunction is unclear, but researchers have identified some genetic variations that may increase a person’s risk.

## There are two general types of the disease:

- **Localized scleroderma** (also known as morphea), which usually affects only the skin and can sometimes go away on its own.
- **Systemic scleroderma**, which usually affects small or large areas of skin. It can also affect internal organs, which can be deadly.

## Who gets it?

An estimated 300,000 Americans have scleroderma (about 100,000 with systemic scleroderma and 200,000 with the localized form). Anyone can get it, but you can’t catch it from other people. It’s most common in adults and especially in women. The localized type is more likely to develop before the age of 40, while the systemic type is more common in adults ages 30 to 50 and in African-Americans.

## What are the symptoms?

Among the most common symptoms are:

- Patches of hard or thickened skin that appear shiny.
- Fingers or toes that turn blue or white or become numb in cold weather, called Raynaud’s syndrome, which is caused by a narrowing of blood vessels.
- Painful bumps under the skin from calcium deposits.
- Red spots on the face or hands from the swelling of tiny blood vessels.
- Digestive problems, including heartburn, acid reflux, and trouble swallowing, from swelling of the esophagus.

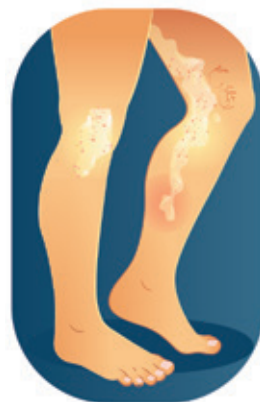
## How is it diagnosed?

There is no single test to diagnose scleroderma, but there are blood tests to detect certain proteins (called antibodies) produced by the immune system, as well as a physical exam, skin biopsy, and lung, heart, and gastrointestinal tests.

## How is it treated?

There are some drugs patients can take to relieve symptoms and reduce damage.

Immunosuppressants, such as mycophenolic acid, have become a major category of medications used against the overactive immune system. Repurposing or combining drugs used for cancer and other diseases have also led to promising results. Patients are encouraged to make changes to their diet, learn stretching to help their joints, and use creams to help with red patches of skin. ■



Scleroderma can lead to painful bumps under the skin, red spots, patches of hard skin, and even digestive problems.

**SOURCES:** National Institute of Arthritis and Musculoskeletal and Skin Diseases; Genetics Home Reference; National Institute of Allergy and Infectious Diseases; NIH RePORT

# 5 Things To Know If You Have Scleroderma

*Maureen Mayes, M.D., is the founder and director of the Scleroderma Clinical and Research Program at the University of Texas Health Science Center, Houston. Her research on scleroderma has been supported by NIH. We asked her for her top advice for those diagnosed with the disease.*

- 1 Don't get discouraged.** "Treatments have improved, so the outlook for patients is more optimistic than it was 16 or 17 years ago," Dr. Mayes says. Unfortunately, some health care providers who aren't aware of these newer developments may give patients a bleaker impression. "I want patients to know they have more choices now."
- 2 Get a second opinion.** Scleroderma is a rare disease and primary care providers or internists who haven't treated many cases of scleroderma may not be as familiar with the latest tests and treatments, or even with the range of often subtle symptoms. There are more than two dozen scleroderma centers across the U.S., with specialists trained in treating the condition. Patients are often hesitant to tell their health care provider they want to get a second opinion, "but getting another viewpoint can be very helpful. At least go for an evaluation," she says.
- 3 Get regular heart and lung testing.** Systemic scleroderma can cause scar tissue buildup in the lungs and heart, so "it's very important that patients get annual or biannual testing so any problems can be identified early and treated appropriately," Dr. Mayes says.
- 4 Get the right diagnostic test.** Trouble getting an accurate diagnosis is a common problem that Dr. Mayes hears from patients. The key is an ANA/IFA blood test, which stands for antinuclear antibody using indirect immunofluorescence. The problem is that a faster, less expensive, simplified ANA test has been introduced, but it can produce false negative results for scleroderma patients. Dr. Mayes urges patients to get the ANA/IFA test.
- 5 Seek tips from other patients.** Local or online support groups and scleroderma organizations are a great source of tips for improving a patient's quality of life, Dr. Mayes says. "Even little things, like always having a jacket and gloves with you to protect Raynaud's-affected fingers from drastic temperature changes in air-conditioned stores and offices, are important." ■



## Struggling for Answers: A Patient’s Scleroderma Journey

Dee Burlile’s children and MedlinePlus helped her through diagnosis

**I**t took Dee Burlile four frustrating, painful years to find out that all the symptoms she had been suffering from were the result of one disease: scleroderma.

It’s a common story for those with this rare condition, which is why Dee now urges patient groups around the country to learn from her experience.

“You must stick up for yourself,” Dee says. “You are your own best advocate and if you’re not getting the help you need, look for other resources. Find people who will listen and help you.”

### The day before Thanksgiving

Dee’s story begins in 2012, the day before Thanksgiving, when she was home in Meridian, Idaho, with her three children. She took a quick shower, walked into her bedroom, and collapsed.

Her 4-year-old son found her and quickly called his 6-year-old sister, who called 911 and then told her older brother to run outside and find a neighbor to help.

“My children literally saved my life,” says Dee, who says she remembers nothing of that morning “until I woke up to EMTs telling me I had been near cardiac arrest.”

Dee had been having strange symptoms for a while, including fingers that turned white in the cold, calcium deposits, joint problems, heart problems, and gastrointestinal issues that had led her to lose 40 pounds.

“All the clues were right there, but none of my doctors seemed to be able put the pieces together.”

When she suggested to one doctor that she might have a condition responsible for all her problems, he told her, “I’m not going to look for zebras if the zoo isn’t in town”—meaning, why look for something unusual if nothing unusual is happening. He also suggested her problems were from the stress of caring for three young children and not eating enough.

### Still searching for clues

After four years of steadily worsening symptoms, she was ultimately sent to a rheumatologist who took a blood test and told her she had scleroderma.

“I didn’t even know what it was,” she remembers. “There were no specialists in our town, no support groups. I felt lost.”

So she searched online on MedlinePlus and PubMed for anything she could find about scleroderma. “I must



Dee Burlile, pictured here with her family, credits her three children with saving her life.

have printed out two trees’ worth of paper with detailed information about my condition that I never knew,” Dee says.

It was a link on MedlinePlus that also led her to the Scleroderma Foundation, where she found support groups and help in getting officially diagnosed at the Mayo Clinic in Minnesota in 2016.

### An updated treatment regimen

She also found scleroderma specialists in Seattle, about eight hours away, who have started her on a regimen that includes some of the newest drugs being used to treat scleroderma’s many complications.

She takes 35 medications and treatments, including weekly immunosuppressant injections, a nitroglycerin ointment, and a compression device to improve blood circulation in her legs. She also credits doing daily yoga stretches from a chair with allowing her to walk without a walker.

Although Dee now advises other scleroderma patients working with the Scleroderma Foundation, it’s health care providers she wishes she could convince to change their attitude about patients with puzzling conditions.

“I wish some doctors would make more of an effort to talk to their patients’ other doctors and do some digging,” Dee says. “Sometimes,” she adds, “a doctor really has to look for a zebra.” ■



# What's New on the Horizon for Scleroderma?

## NIH-supported experts review stem cell transplantations, anti-fibrosis drugs, and more

There may not be a cure yet, but many scleroderma experts are optimistic about ongoing research into new treatments.

We asked John Varga, M.D., professor of medicine and director of the Northwestern University Scleroderma Program, and Carol Feghali-Bostwick, Ph.D., a scleroderma patient herself and a professor of medicine at the Medical University of South Carolina, to discuss potential new advancements. Both researchers work with NIH's National Institute of Arthritis and Musculoskeletal and Skin Diseases.

“Stem cell transplants could be a promising treatment for those with very severe scleroderma.”

- John Varga, M.D.



Carol Feghali-Bostwick, Ph.D. (left) and John Varga, M.D. (right) work with NIH on scleroderma research.

**Anti-fibrosis drugs:** There are major lung studies looking at drugs that can halt the development of thick scarring tissue, called fibrosis, that can occur in systemic scleroderma, as well as other lung diseases like asthma and chronic obstructive pulmonary disease, Dr. Feghali-Bostwick says.

“My team and I also have used NIH funding to identify an essential peptide—a small piece of protein—that seems to be able to stop and possibly reverse the formation of fibrosis in mouse and human tissue. We are now researching how this peptide works, with hopes of testing it in a clinical trial,” she adds.

**More drugs for rare diseases:** Many immunomodulatory drugs, which can regulate the immune system, are in clinical trials. And a number of recent studies have found that drugs that block an inflammation-causing protein in lupus and similar diseases may also work for scleroderma.

**Stem cell transplants:** There is a lot of interest in a recent NIH-funded study that found that stem cell transplants for people with severe scleroderma could have

long-term, beneficial results, says Dr. Varga. In the study, chemotherapy and radiation were used to wipe out the person's immune system, and then the person's own stem cells were used to rebuild a new system. But Dr. Varga and others warn that a stem cell transplant is not appropriate for every patient and can involve dangerous side effects. “But it could be a promising treatment for those with very severe scleroderma,” he says. ■

**SOURCE:** NIH News Releases

### Find Out **More**

- ▶ **MedlinePlus: Scleroderma**  
<https://medlineplus.gov/scleroderma.html>
- ▶ **National Institute of Arthritis and Musculoskeletal and Skin Diseases: Scleroderma**  
<https://www.niams.nih.gov/health-topics/scleroderma>
- ▶ **Scleroderma Research Foundation**  
<http://www.srfcure.org/>
- ▶ **Scleroderma Foundation**  
<http://www.scleroderma.org/>

# FIGHTING the Opioid CRISIS:

## NIH HEAL Initiative Takes on Addiction and Pain Management

**M**ore than 115 Americans die from opioid overdose every day. That's four times higher than the number of opioid overdose deaths that occurred 20 years ago.

It is now a major public health crisis in the United States. Congress doubled funding for opioid abuse and addiction research at the National Institutes for Health (NIH) to nearly \$1.1 billion in 2018.

### Launching HEAL

In April 2018, NIH launched a research initiative called the NIH HEAL (Helping to End Addiction Long-term) Initiative.

Government agencies, including NIH, are focused on finding new ways to combat the opioid crisis. HEAL researchers will aim to improve treatments for opioid misuse and addiction, and enhance pain management.

"We've learned a lot about the neuroscience of pain and what areas we can address that have not been fully addressed so far," says NIH Director Francis S. Collins, M.D., Ph.D. "With the national opioid tragedy we are facing, we can do better with partnerships with the Food and Drug Administration (FDA) and other research agencies."

### Opioid crisis: From bad to worse

How did the opioid crisis begin?

One common theory among scientists is that the opioid crisis began in the 1990s because health care providers overprescribed opioid medications to treat pain.

"Over the course of 20 years, opioid addiction became more and more prominent," Dr. Collins says. "As people became dependent on opioid drugs, they needed to

continue taking them to avoid withdrawal symptoms, and needed higher doses to achieve the feelings they were seeking."

Meanwhile, the supply of heroin, a type of opioid that's illegal in the U.S., became more accessible and affordable. Because of this, some people started using heroin instead of things like prescription opioids, because it is cheaper and easier to get.

"Tragically, synthetic opioids that are 100 times more potent than heroin have entered the mix, and people nationwide are putting themselves at risk," says Dr. Collins.

Heroin is naturally derived from opioid plants, but now there are synthetic opioids that are more common—and more fatal.

### A community approach

One of the main NIH projects that will be funded through HEAL is the HEALing Communities Study.

Launching in 2019, its goal is to address the opioid crisis by improving treatments for pain and addiction and reduce deaths caused by opioid overdose.



NIH Director Francis Collins, M.D., Ph.D.

“The experience of addiction is a disease. It’s associated with changes in the brain that cannot simply be willed away by a motivated person.”

– NIH Director Francis Collins, M.D., Ph.D.



“We want to address the needs of patients suffering from pain and do it responsibly,” says Nora Volkow, M.D., director of the National Institute on Drug Abuse at NIH. “Currently, the way we measure pain is on a scale of one to 10 with smiley faces. That may tell us how intense your pain is, but it is not predictive of success.”



**Nora Volkow, M.D., is the director of the National Institute on Drug Abuse, which is researching new ways to treat pain and help those with addiction.**

Dr. Volkow is researching new types of drugs that address pain and studies aimed at improving access to high-quality, evidence-based addiction treatment programs. The programs will be tested across several community-based systems such as health care, mental health, and criminal justice systems to test the effect of medication-assisted treatment, long-term treatment, and more access to recovery support services.

“For example, we have models in prison to treat people who are addicted, but we want to be sure they have care once they leave prison, before they return to the community,” Dr. Volkow says.

### Understanding pain

Since pain is at the root of the opioid crisis, HEAL will explore new, nonaddictive ways to treat the nearly 25 million Americans suffering from daily chronic pain.

That includes researching how light pain develops into chronic pain, how chronic pain makes people susceptible to opioid misuse, and why some people have chronic pain and others don't.

The initiative will research the biology of pain, hold clinical trials, and work with pharmaceutical companies and the FDA to develop better nonaddictive pain management treatment.

### Reframing addiction

In addition to researching opioid addiction and pain management, NIH and the broader medical community are working to reframe our conversation about opioids. That starts with removing the stigma around addiction.

“The experience of addiction is a disease,” says Dr. Collins. “It’s associated with changes in the brain that cannot simply be willed away by a motivated person.”

People who have addictions need treatment, he adds, noting that they must be considered as people who have fallen into this situation and are in need of help.

“Don’t stigmatize them,” Dr. Collins adds. “Otherwise we will never make it possible for those people to get the help that they need.” ■

SOURCE: NIH HEAL Initiative

# The Opioid Crisis: An Overview

## Who is affected?

Roughly **2 million people** live with an opioid addiction in the U.S.

“It’s important to note that people typically think about individuals who are addicted to opioids as stereotypes,” says NIH Director Francis Collins, M.D., Ph.D. “In reality, the type of affected individuals is very broad; **there are people in rural and urban communities and all socio-economic classes**, and sometimes even multiple generations of families.”

That includes babies, teenagers, parents, and grandparents, **all genders, and all ages**.

The next generation of newborn babies is a serious concern, as women who are addicted to opioids while pregnant may not share this information, for fear that their newborn babies could be taken away.

## What is opioid addiction or misuse?

**Opioids** are a class of drugs that include illegal drugs like heroin, synthetic opioids like fentanyl (which can be obtained legally and illegally), and pain relievers available legally by prescription like oxycodone, codeine, and morphine.

**An opioid addiction** is a chronic brain disease. It causes a person to compulsively seek out drugs, despite the negative consequences.

**Opioid misuse** means that you aren’t using a drug as medically intended, such as when a person does not take medicine according to the prescription instructions or a person takes an illegal opioid. ■

**SOURCES:** National Institute on Drug Abuse; Substance Abuse and Mental Health Service Administration’s National Helpline

## Healing and Haiku:

### One Parent’s Experience with Addiction and Loss

Haiku is a form of poetry that can express complex emotions and ideas in a short and simple form.

In his book, “Haiku Snapshots: Reflections on Drug Addiction,” Wally Berger shares haikus detailing the experience of losing a child to opioid addiction. The book is written across the lifetime of his son and recounts both happy and difficult times.

Below are a few examples.

so sweet a baby	:	at night he sleeps light	:	he’s a good person
no early signs of problem	:	guarding his money and life	:	drug addiction not addict
high expectations	:	living on the edge	:	not disease defined

# 1-800-662-HELP (4357)

If you or a loved one needs help with opioid misuse or other types of addiction, call the Substance Abuse and Mental Health Service Administration’s National Helpline at **1-800-662-HELP (4357)**. It is free, confidential, and available 24/7, 365 days a year.

# Renewal and Recovery after Opioid Dependence

*Recovering addict gets support from nonprofit youth-at-risk program*

**F**or the past six years, James M. was in and out of jail, courtrooms, detox, and halfway houses.

“I started using marijuana when I was 11 years old,” James says.

When he was 18, James started trying other drugs, including cocaine and the prescription opioid oxycodone, which is used to treat moderate to severe pain. Both drugs can be highly addictive.

James said he was often desperate and would try any opioid he could get.

“I tried to stop,” he says, “but my body would be achy, and I’d feel sick. So, I just kept doing more.”

Now, 25-year-old James has been drug free for more than a year.

He has been working and taking classes to get his high school equivalency certificate, thanks in part to his involvement with UTEC, Inc. This nonprofit program in Lowell, Massachusetts, works specifically with young adults who have proven risk factors.

## Working with UTEC

UTEC’s mission is to trade violence and poverty for social and economic success. They nurture and train young people who have been in prison or struggled with violence and gang involvement. Many of these young adults (ages 17-25) have histories of or current issues with drug use, including opioids.

UTEC gives these young people a chance to stay out of trouble and to work, earn a high school equivalency certificate, and learn skills such as woodworking or cooking. Most of all, UTEC helps teach how to be

responsible, stay clean, and show up for work every day.

“I first talked to UTEC when I was 19,” James says. “They came to my jail, and asked me if I wanted to be involved. I tried the program, but I couldn’t stick with it.”

James would find himself back in jail, then in mandatory detox centers and therapy, but eventually he’d turn back to drugs.

## Ready for responsibility

James ultimately grew tired of going to halfway houses, detox centers, and jail.

“I was sick of it all,” he says.

When UTEC approached him in jail again in the summer of 2018, James was ready. He understood that UTEC could help him stay away from drugs while learning job skills.

James has been coming to UTEC regularly for more than five months. He is getting paid for the work he does at UTEC, including cooking and helping others.

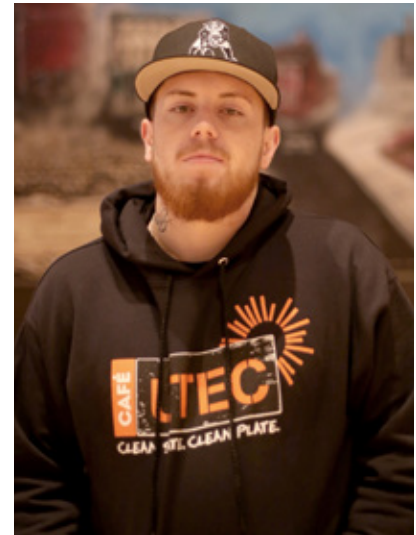
## A rough road

James admits that the UTEC program wasn’t easy. If James didn’t show up for work, a UTEC member would come to his house and make sure he was OK.

“Sometimes I wouldn’t even open the door. I didn’t want to talk to them. A lot of people in this program are addicts or gang members. UTEC will do a lot to get you to come back,” he says. “They’re really supportive.”

At first, James didn’t attend the optional extra class at the end of the day.

“I would just walk home or have my girlfriend pick me up,” he says, “but then I realized it kept me busy,



James M. has been drug-free for more than five months after working with UTEC, Inc.

and the better attendance I had, the better jobs I’d get.” He has enjoyed programs such as UTEC’s outdoor leadership hiking program.

## Sobriety and support

James is determined to stay drug free and keep up his attendance at UTEC.

“Drugs are always on the back of my mind, but I keep busy and I have good support,” James says.

Currently, James, his girlfriend, and their baby boy live in his mother’s house. His goal is to find a good job so his family can live on their own.

James is motivated by more than just his own experience.

His cousin died from cocaine laced with fentanyl (a deadly synthetic opioid) while James was in jail, and James’ father overdosed when James was 2 years old.

“That made me see it all. I don’t want to do that to my son,” he says.

For people who know an addict, James offers this advice: “Don’t attack or accuse them—it only makes it harder for them. If you can’t help an addict, don’t hurt one.” ■

# A Safe Home for Young Men Struggling with Addiction

*Parents and community build sober home for recovering opioid addicts*

**B**arbara Gillmeister knows what it's like to worry about a child with an opioid addiction. "When you have someone who is actively using [drugs], you're always waiting for that phone call," she recalls.

Unfortunately, Barbara and her husband David did get a devastating phone call. Their son, Steven, known as "Gilly" to his friends, died of a heroin overdose at the age of 25, after struggling with drug addiction for years.

"Steven had a warm heart. He lit up a room with his smile," Barbara says. "He was so generous and made everyone, no matter who walked through the door, feel welcome."

After Steven passed away, the Gillmeisters opened a residential home, or "sober house." Gilly's House is for young men who have struggled with addiction and want to continue their recovery.

Thanks to donations from family and friends, they opened Gilly's House with support from the SAFE Coalition, a regional coalition of community partners in Western Norfolk County, Massachusetts.

Gilly's House offers a stable and safe environment for newly sober young men in their 20s or 30s who are in recovery. Residents participate in self-help groups and 12-step group meetings, and are assisted with setting and achieving goals to help transition to community life.

"Our goal is to have these young men leave here with the tools they need to make it on their own," Barbara says.

From cooking, to paying bills, to light carpentry, cleaning, and other life skills, Gilly's House also offers opportunities for social, educational, and vocational growth.

A daily structured schedule reinforces a lifestyle free of alcohol and drug use.

"Once you are sober, if you don't have skills to take care of yourself when you leave Gilly's House, you are apt to fall back to old patterns," Barbara says.

Each resident must be sober for at least 40 days, employed, and follow strict house rules, including a curfew.

The young men receive a furnished bedroom, toiletries, and a fully stocked kitchen to cook their own meals.



Top, Barbara Gillmeister and her son Gilly. Right, a sign for Gilly's House.



**"If I can help a few people, that makes all the work we've done worthwhile."**

– Barbara Gillmeister

Almost all of the amenities are donated or funded by families of opioid addicts and friends.

"It seems that everyone has experienced a loved one or someone close in their lives with drug addiction, and so many people give back in the most amazing ways," Barbara says.

After Gilly passed away, Barbara thought she would never have anything to do with drugs and addiction.

"Now I am surrounded by people who have lost children and want to help others," she says. "It's overwhelming. If I can help a few people, that makes all the work we've done worthwhile." ■

# Acting Now to Help Newborns with Opioid Withdrawal

*NIH study helps mothers and their children*

**A**t the Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD), Director Diana Bianchi, M.D., is working with a team of researchers to address issues associated with neonatal opioid withdrawal syndrome, also called neonatal abstinence syndrome.

The condition happens in babies exposed to opioids in the womb, then abruptly removed from drug exposure when they are born.

The Centers for Disease Control and Prevention says that the number of neonatal opioid cases increased approximately 300 percent from 1999 to 2013.

Many newborns who are exposed to opioids before they are born have difficulty eating and sleeping and are hard to calm down.

They may also be at higher risk of developmental, behavioral, and educational problems—and possible use of opioids and other substances during their own reproductive years.

NICHD has teamed with NIH's Environmental Influences on Child Health Outcomes Program to launch a series of studies to address the specific medical and social needs of these infants.

The project is called Advancing Clinical Trials in Neonatal Opioid Withdrawal Syndrome, or ACT NOW.

The goal of the project is to help providers learn how to best treat these infants. ACT NOW researchers are working across 20 sites in the U.S.

Soon these efforts will expand into large clinical trials and follow-up studies to provide evidence that will improve how providers identify and



**“It’s a major public health problem. We’re trying to break the intergenerational drug dependency.”**

**—Diana Bianchi, M.D.**

treat babies with opioid withdrawal.

“Most of the information we have on opioid exposure is from moms who used heroin and methadone,” says Dr. Bianchi. “Now we have newly developed synthetic narcotics, and we have no idea what effects they have on the brain.”

Women of reproductive age, whether or not they become pregnant, are also at risk of developing opioid use disorders.

Conditions such as endometriosis, ovarian cysts, and pelvic inflammatory disease are very common, and women with these conditions often have chronic pain requiring prescription pain medications.

“ACT NOW starts at the end of the problem,” says Dr. Bianchi. “Our efforts need to also focus on going back to early reproductive health issues and examine why women are getting opioids, how can we educate providers, and what can we do at a national level to influence that.”

One of the many goals for NICHD is for infants and mothers to stay in contact with researchers so that the long-term effects of exposure to opioids can be identified.

NICHD is especially interested in the brain development of babies exposed to opioids in the womb.

“It’s a major public health problem,” says Dr. Bianchi. “We’re trying to break the intergenerational drug dependency.” ■

## Find Out **More**

► **MedlinePlus: Opioid Abuse and Addiction**

<https://medlineplus.gov/opioidabuseandaddiction.html>


► **National Institute on Drug Abuse: Opioids**

<https://www.drugabuse.gov/drugs-abuse/opioids>

► **NIH HEAL Initiative**

<https://www.nih.gov/research-training/medical-research-initiatives/heal-initiative>

# Using Your Own Immune System to Fight Cancer



NIH experimental immunotherapy work is delivering promising, personalized results for some cancer patients

*For more than 30 years, Steven A. Rosenberg, M.D., Ph.D., has pioneered the development of immunotherapies and gene therapies for patients with advanced cancers. These therapies use patients' own bodies to fight cancer. Dr. Rosenberg is chief of surgery and senior investigator at the National Cancer Institute (NCI). He spoke to NIH MedlinePlus magazine about how a new experimental treatment has shown success for patients in a clinical trial.*

## What should readers understand about treating cancer with immunotherapy?

Up to now, we have had three major ways to treat cancer—surgery, radiation therapy, and chemotherapy. But the number of patients who develop cancer is so high that the best application of those three treatments in 2017 still resulted in 600,000 cancer deaths in the U.S. We've been developing a fourth approach to cancer, with increasing effectiveness: immunotherapy.

The body's own immune system recognizes cancer as foreign, much as it would a virus. And the immune system can eliminate a virus. It turns out that the body can recognize the cancer as foreign but not foreign

enough to eliminate it. Our goal in immunotherapy is to stimulate the body's own natural defenses to fight the cancer.

There has been some startling progress made in the last several years. Immunotherapy has helped patients who can't be effectively treated by conventional surgery, radiation, and chemotherapy.

### FAST FACT

In 2016, there were an estimated **15.5 million cancer survivors in the U.S.** That number is expected to increase to **20.3 million by 2026.**





Steven Rosenberg, M.D., Ph.D., in his lab at the National Cancer Institute.

### What is driving this approach?

A lot of basic biologic research has been translated into effective treatments. That's the way that science works. We learn from basic research. When we have enough information, we can apply that information.

Right now, my own involvement is in the application of basic research findings from my own laboratory, applied to the treatment of patients.

### How is your current research translating into treatments?

We developed the clinical application of Interleukin-2. This treatment helps strengthen a patient's immune system to fight off cancer. It was the first immunotherapy approved by the Food and Drug Administration for patients with metastatic melanoma [a type of skin cancer] and

**“Our goal in immunotherapy is to stimulate the body's own natural defenses to fight the cancer.”**

– Steven Rosenberg, M.D., Ph.D.

metastatic kidney cancer. Metastatic cancer is cancer that has spread throughout the body.

Along with Interleukin-2, we have taken a patient's own immune cells, selected those cells that attack the cancer, and then give them back into the patient. That combination appears to be capable of treating almost 50 percent of these patients, even though other treatments were not successful.

We have seen some responses in patients with colon cancer, liver cancer, and recently with breast cancer. That is one of the most exciting areas of immunology research.

### Are you still recruiting patients for this or any other trials that are related to immunotherapy?

Yes. We are looking for patients who have metastatic solid cancers [cancers in organs] of the gastrointestinal tract or ovarian cancer who have been through standard treatments that have not worked. We want to apply our new treatments in a variety of clinical trials.

Our referral office can be reached by calling 1-866-820-4505 or emailing [IRC@nih.gov](mailto:IRC@nih.gov). ■

# CLINICAL TRIAL

## Gives Breast Cancer Patient *a Second Chance*

*Could Judy Perkins' own immune system help fight her cancer?*

**W**hen Judy Perkins got the diagnosis of stage IV metastatic breast cancer, or breast cancer that has spread, she felt defeated.

"It was pretty much a death sentence," she says now. "I thought I knew all the treatments, and I had run out of treatments."

It was Judy's diagnosis that led her to become active in the National Breast Cancer Coalition and advocate for other breast cancer patients. "In July 2015, I took a class designed to bring breast cancer advocates up to speed on all the science," Judy said.

The class is where she met Stephanie L. Goff, M.D., a member of a pioneering cancer research team at NIH. Dr. Goff works with Steven A. Rosenberg, M.D., Ph.D., of the National Cancer Institute (NCI) in Bethesda, Maryland.

Dr. Goff mentioned to Judy that the surgery branch of NCI was running a clinical trial on immunotherapy at NIH. It sounded like it might be a good match for Judy's case. Judy had already participated in clinical trials before with mixed success, but she was open to another chance.

Following their meeting, Judy participated in a screening test with Dr. Goff and Dr. Rosenberg and was later accepted into the trial.

### **A chance with a clinical trial**

To treat Judy, Dr. Goff and her researchers sequenced (or studied the structure of) the DNA and RNA in one of her cancerous breast tumors and in her normal tissue. They wanted to see which mutations were unique to her cancer.

The procedures took place at the NIH Clinical Center hospital in Bethesda.

The researchers tested different cells that specifically target tumor mutations to see if they could shrink tumors. They then put those anti-tumor cells back into Judy's body.

Then as part of the immunotherapy treatment, Judy underwent more chemotherapy and had a catheter-like port inserted into her body to infuse the anti-tumor cells.

*"I knew within 10 days that things were moving in the right direction."*

– Judy Perkins

"We took the lymphocytes (white blood cells) that were in Judy's body—the warriors of her immune system—that are constantly circulating throughout her body," says Dr. Rosenberg, who supervised the trial. "We especially wanted the tiny number of lymphocytes that recognized the mutations in her cancerous tumors. And we increased the number of those cells to treat Judy with her own anti-tumor cells. This is, to me, one of the most exciting areas of immunology research."

Judy knew right away that this treatment was different.



Judy Perkins on a recent vacation (left) and receiving an infusion of tumor-infiltrating lymphocytes as part of her cancer treatment (right).

### A second chance

“I knew within 10 days that things were moving in the right direction,” Judy said. “Before I left the Clinical Center around Christmas 2015, I knew that the tumor was starting to get a little softer and was shrinking. But I didn’t know whether it had to do with the infusion. It could have just been from the chemo. I knew it was a positive thing. I just didn’t know if it was going to continue.”

But it did continue. The tumor had dissolved, along with her other tumors. “My energy had come back,” said Judy. “I knew that this was a way different response from anything else I’d had. This was big.”

### FAST FACT

The five most common cancers in the U.S. are breast cancer, lung and bronchus cancer, prostate cancer, colon and rectum cancer, and melanoma.

It was the best Christmas gift of Judy’s life. She has felt better ever since that winter in 2015.

### Find the magic

Judy urges other cancer patients to explore clinical trials and see if they may be the right fit for their condition.

“At first, I wanted to ‘find some magic’ for myself,” Judy says. “Later, as I got sicker and less hopeful about ‘finding the magic,’ I figured that it might do some good for people coming after me.”

She recommends that people speak with their health care provider to make the best decision for their case and that they speak up immediately if they have any concerns or questions during a trial.

“If you participate, you should be aware that if something doesn’t feel right, you should discuss this immediately with your doctor,” Judy says. “Sometimes the side effects are unknown and you really need to pay attention and speak up.

My philosophy for all of my treatments was to prepare for the worst and hope for the best.”

Judy ended up “finding some magic” after all. After three years, she remains cancer free. ■

### Find Out More

- ▶ **MedlinePlus: Immunotherapy**  
<https://medlineplus.gov/cancerimmunotherapy.html>
- ▶ **MedlinePlus: Breast Cancer**  
[www.ncbi.nlm.nih.gov/gtr/](http://www.ncbi.nlm.nih.gov/gtr/)
- ▶ **National Cancer Institute: Breast Cancer**  
<https://www.cancer.gov/types/breast>
- ▶ **National Cancer Institute: Immunotherapy**  
<https://www.cancer.gov/about-cancer/treatment/types/immunotherapy?redirect=true>

from  
the

# lab

LATEST  
RESEARCH  
UPDATES  
FROM NIH

## High Blood Glucose During Pregnancy Increases Chance of Mother's Type 2 Diabetes, Child's Obesity

**A MOTHER'S HEALTH DURING PREGNANCY CAN HAVE A LASTING IMPACT** on both her health and that of her child.

A new NIH study found that pregnant women with higher than normal blood glucose (blood sugar) are more likely to develop type 2 diabetes later in life. This was found even in moms who hadn't been diagnosed with gestational diabetes (diabetes during pregnancy).

Also, children of mothers with higher than normal blood glucose

during pregnancy were more likely to be obese.

Type 2 diabetes occurs when your body stops using or producing insulin well. It is preventable, unlike type 1 diabetes, which people develop when their bodies' immune systems attack the cells that make insulin.

The National Institute of Diabetes and Digestive and Kidney Diseases led funding of the study, which checked in with mothers and their children 10 to 14 years after birth. The study is a follow-up to one conducted during

the same mothers' pregnancies and shortly after birth.

Among the women with higher than normal blood glucose during pregnancy, nearly 11 percent had type 2 diabetes at the follow-up study visit. Forty two percent had prediabetes, which means blood glucose levels are high but not high enough to be diagnosed as diabetes.

In comparison, among those without higher than normal blood glucose during pregnancy, about 2 percent had type 2 diabetes and about 18 percent had prediabetes.

What can pregnant moms do?

Three things:

- Stay active
- Maintain healthy blood pressure
- Eat healthy foods

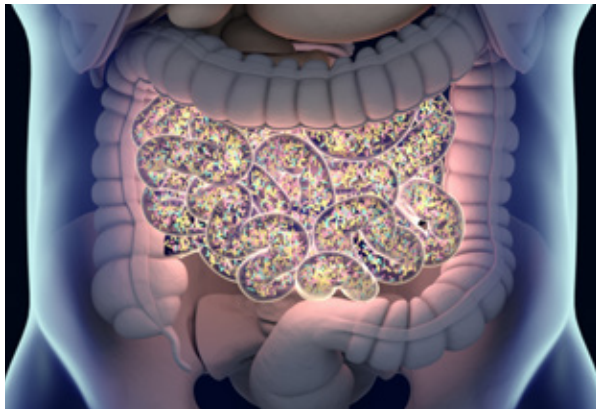
By doing these three things, moms can help lower their risk of developing higher than normal blood glucose during pregnancy and type 2 diabetes afterward.

Additional support was provided by the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development. ■

**SOURCES:** NIH News Releases; National Institute of Diabetes and Digestive and Kidney Diseases



IMAGE: ADOBE STOCK



## Restoring Beneficial Gut Bacteria in Cancer Patients

### COULD OUR OWN BACTERIA BE HELPFUL IN CANCER TREATMENT?

Maybe, say researchers sponsored in part by the National Institute of Allergy and Infectious Diseases (NIAID).

Antibiotics are essential to prevent life-threatening and serious bacterial infections in patients undergoing cancer treatment. However, antibiotics also destroy “good” bacteria that prevent infections and enhance immune function.

Fecal microbiota transplantation, also known as FMT, is a new way to replenish “good” bacteria in cancer patients who require stem cell transplants, researchers found.

The process involved using a patient’s own stool, known as autologous fecal microbiota transplantation, or auto-FMT for short. Before their stem cell transplantation, the patient’s stool was frozen and reintroduced after the stem cell transplant was stable, with the objective of restoring good bacteria to their gut.

While the process might sound strange, it had promising results.

In the study, auto-FMT resulted in the recovery of beneficial gut bacteria to near-normal levels within days for patients. Under more traditional treatment, it can take several weeks for good bacteria to return to normal levels.

“This important study suggests that clinical intervention using auto-FMT can safely reverse the disruptive effects of broad-spectrum antibiotic treatment,” says NIAID Director Anthony S. Fauci, M.D. “If validated in larger studies, this approach may prove to be a relatively simple way to quickly restore a person’s healthy microbiome.” ■

SOURCE: NIH News Releases

## A Wearable Blood Pressure Monitor May Be in Our Future

**MORE AND MORE CONSUMERS ARE USING HEALTH MONITORS** they can wear. These wearables make it easier for patients to track their own health on a daily basis and stay motivated.

Soon, blood pressure may be something patients can track with wearables 24/7. NIH-supported researchers are testing a new wearable that can monitor a patient’s blood pressure with a small, wearable skin patch.

The patch monitors a patient’s blood pressure more accurately than other methods, like an inflatable cuff around the arm. This is because it’s able to record blood pressure in the central arteries and veins, which are located in our necks.

The patch’s ultrasound waves monitor subtle, real-time changes in the shape and size of pulsing blood vessels. These changes indicate when pressure rises and drops.

The sensor can monitor pulses more than an inch beneath the skin. That’s deeper than previous patches and other skin sensors.

NIH Director Francis H. Collins, M.D., Ph.D., said the new technology shows great promise. “So far, the new device appears to function better than any commercially available, noninvasive device for measuring central blood pressure,” he wrote in his blog.

Currently, the patch needs to be connected to wires to monitor central blood pressure, but the researchers hope to develop a wireless version.

“The hope is that one day soon their device will offer round-the-clock monitoring of central blood pressure. That could utterly transform our management of hypertension,” Dr. Collins added. ■

SOURCE: NIH Research Matters



NIH-supported researchers are testing a wearable blood pressure monitor. They hope to develop a wireless version, like the one seen here, soon.

# NIH on the web

## Could Llamas Give Us Clues about the Flu?

➔➔ **Researchers are exploring** how these furry animals can help us better understand how to fight off the flu.

Why llamas? The South American animals have smaller antibodies (which protect against viruses) than humans. Because they're smaller, they're able to reach parts of the flu virus that bulkier human antibodies can't.

Using llama antibodies in mice, researchers are testing how new gene therapies, specifically a "gene mist," may fend off a range of flu viruses better than other methods.

NIH Director Francis Collins, M.D., Ph.D, discusses this early research in a post on his Director's Blog. ■

**SOURCE:** NIH Director's Blog



Find it all in one place!  
[magazine.medlineplus.gov](https://magazine.medlineplus.gov)



## New MedlinePlus Video Explores How to Counter Opioid Overdose

➔➔ **More than 115 people in the U.S.** die every day after overdosing on opioids. One important tool for helping opioid overdose is naloxone, a drug that helps reverse the effects of an overdose.

A new animated video from MedlinePlus and the National Institute on Drug Abuse explains just how naloxone works in our bodies to counter the effects of opioid overdoses.

The video will be available in Spanish and English, and can be found on the National Library of Medicine's YouTube channel. ■



## Out with the Cookies, in with the Veggies!

➔➔ **As you take steps to eat healthier** as the new year begins (and hopefully all year-round), NIH is here to help.

Check out free heart-healthy recipes from NIH's National Heart, Lung, and Blood Institute (NHLBI), which are low in saturated fat and sodium. They use small amounts of vegetable oil and feature lean meats, low-fat dairy, veggies, fruits, whole grains, beans, and nuts.

From Caribbean casserole to tilapia with tomatoes, there are more than 180 dishes available, including desserts! You can access 40 recipes on MedlinePlus.gov in both Spanish and English. All 180 dishes are available on NHLBI's website.

Start your new year by eating healthy with fresh herbs, lots of spices, and NIH.

**SOURCES:** Deliciously Healthy Eating: <https://healthyeating.nhlbi.nih.gov/>; MedlinePlus Recipes: <https://medlineplus.gov/recipes/> ■

# NIH Is Here to Help

**The National Institutes of Health (NIH)**—the nation’s medical research agency—includes 30 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](http://www.nih.gov).

## Institutes

### National Library of Medicine (NLM)

[www.nlm.nih.gov](http://www.nlm.nih.gov)  
888-FIND-NLM 888-346-3656

### National Cancer Institute (NCI)

[www.cancer.gov](http://www.cancer.gov)  
800-4-CANCER 800-422-6237

### National Eye Institute (NEI)

[www.nei.nih.gov](http://www.nei.nih.gov)  
301-496-5248

### National Heart, Lung, and Blood Institute (NHLBI)

[www.nhlbi.nih.gov](http://www.nhlbi.nih.gov)  
301-592-8573

### National Human Genome Research Institute (NHGRI)

[www.genome.gov](http://www.genome.gov)  
301-402-0911

### National Institute on Aging (NIA)

[www.nia.nih.gov](http://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](http://www.niaaa.nih.gov)  
301-443-3860

### National Institute of Allergy and Infectious Diseases (NIAID)

[www.niaid.nih.gov](http://www.niaid.nih.gov)  
301-496-5717

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

[www.niams.nih.gov](http://www.niams.nih.gov)  
877-22-NIAMS 877-226-4267

### National Institute of Biomedical Imaging and Bioengineering (NIBIB)

[www.nibib.nih.gov](http://www.nibib.nih.gov)  
301-451-6772

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

[www.nichd.nih.gov](http://www.nichd.nih.gov)  
800-370-2943

### National Institute on Deafness and Other Communication Disorders (NIDCD)

[www.nidcd.nih.gov](http://www.nidcd.nih.gov)  
800-241-1044 (voice)  
800-241-1055 (TTY)

### National Institute of Dental and Craniofacial Research (NIDCR)

[www.nidcr.nih.gov](http://www.nidcr.nih.gov)  
301-480-4098

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

[www.niddk.nih.gov](http://www.niddk.nih.gov)  
NIDDK Health Information Center  
1-800-860-8747

### National Institute on Drug Abuse (NIDA)

[www.nida.nih.gov](http://www.nida.nih.gov)  
301-443-1124

### National Institute of Environmental Health Sciences (NIEHS)

[www.niehs.nih.gov](http://www.niehs.nih.gov)  
919-541-3345

### National Institute of General Medical Sciences (NIGMS)

[www.nigms.nih.gov](http://www.nigms.nih.gov)  
301-496-7301

### National Institute of Mental Health (NIMH)

[www.nimh.nih.gov](http://www.nimh.nih.gov)  
866-615-6464

### National Institute on Minority Health and Health Disparities (NIMHD)

[www.nimhd.nih.gov](http://www.nimhd.nih.gov)  
301-402-1366

### National Institute of Neurological Disorders and Stroke (NINDS)

[www.ninds.nih.gov](http://www.ninds.nih.gov)  
800-352-9424

### National Institute of Nursing Research (NINR)

[www.ninr.nih.gov](http://www.ninr.nih.gov)  
301-496-0207

## Centers & Offices

### Fogarty International Center (FIC)

[www.fic.nih.gov](http://www.fic.nih.gov)  
301-402-8614

### National Center for Complementary and Integrative Health (NCCIH)

[www.nccih.nih.gov](http://www.nccih.nih.gov)  
888-644-6226

### National Center for Advancing Translational Sciences (NCATS)

[www.ncats.nih.gov](http://www.ncats.nih.gov)  
301-435-0888

### NIH Clinical Center (CC)

[clinicalcenter.nih.gov](http://clinicalcenter.nih.gov)  
301-496-2563

### Office of AIDS Research (OAR)

[www.oar.nih.gov](http://www.oar.nih.gov)  
301-496-0357

### Office of Behavioral and Social Sciences Research (OBSSR)

[www.obsr.od.nih.gov](http://www.obsr.od.nih.gov)  
301-402-1146

### Office of Rare Diseases Research (ORDR)

[www.rarediseases.info.nih.gov](http://www.rarediseases.info.nih.gov)

### Genetic and Rare Disease Information Center

888-205-2311

### Office of Research on Women’s Health (ORWH)

[www.orwh.od.nih.gov](http://www.orwh.od.nih.gov)  
301-402-1770



## What's Cooking?

Every week, MedlinePlus.gov posts tasty recipes approved by the **National Heart, Lung, and Blood Institute**. The meals are limited in saturated fat and sodium, but not in flavor.

**Start your new year off the healthy way. Try one today.**

<https://medlineplus.gov/recipes/>

<https://medlineplus.gov/spanish/recetas/>