

# NIH News in Health

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## Personalized Medicine Matching Treatments to Your Genes

You're one of a kind. It's not just your eyes, smile, and personality. Your health, risk for disease, and the ways you respond to medicines are also unique. Medicines that work well for some people may not help you at all. They might even cause problems. Wouldn't it be nice if treatments and preventive care could be designed just for you?

The careful matching of your biology to your medical care is known as personalized medicine. It's already being used by health care providers nationwide.

The story of personalized medicine begins with the unique set of genes you inherited from your parents. Genes are stretches of DNA that serve as a sort of instruction manual telling your body how to make the proteins and perform the other tasks that your body needs. These genetic instructions are written in varying patterns of only 4 different chemical "letters," or bases.

The same genes often differ slightly between people. Bases may be switched, missing, or added here and there. Most of these variations have no effect on your health. But some can create unusual proteins that might boost your risk for certain diseases. Some variants can affect how well a medicine works in your body. Or they might cause a medicine to have different side effects in you than in someone else.

The study of how genes affect the way medicines work in your body is called pharmacogenomics.



"If doctors know your genes, they can predict drug response and incorporate this information into the medical decisions they make," says Dr. Rochelle Long, a pharmacogenomics expert at NIH.

It's becoming more common for doctors to test for gene variants before prescribing certain drugs. For example, children with leukemia might get the *TPMT* gene test to help doctors choose the right dosage of medicine to prevent toxic side effects. Some HIV-infected patients are severely allergic to treatment drugs, and genetic tests can help identify who can safely take the medicines. "By screening to know who shouldn't get certain drugs, we can prevent life-threatening side effects," Long says.

Pharmacogenomics is also being used for cancer treatment. Some breast cancer drugs only work in women with particular genetic variations. If testing shows patients with advanced melanoma (skin cancer) have certain variants, 2 new approved drugs can treat them.

Even one of the oldest and most common drugs, aspirin, can have varying effects based on your genes. Millions of people take a daily aspirin to lower their risk for heart attack and stroke. Aspirin helps by preventing blood clots that could clog arteries. But aspirin doesn't reduce heart disease risk in everyone.

NIH-funded researchers recently identified a set of genes with unique

activity patterns that can help assess whether someone will benefit from taking aspirin for heart health. Scientists are now working to develop a standardized test for use in daily practice. If doctors can tell that aspirin won't work in certain patients, they can try different treatments.

One NIH-funded research team studied a different clot-fighting drug known as clopidogrel (Plavix). It's often prescribed for people at risk for heart attack or stroke. Led by Dr. Alan Shuldiner at the University of Maryland School of Medicine, the team examined people in an Amish community. Isolated communities like this have less genetic diversity than the general population, which

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can make it easier to study the effects of genes. But as in the general population, some Amish people have risk factors, such as eating a high-fat diet, that raise their risk for heart disease.

Many of the Amish people studied had a particular gene variant that made them less responsive to clopidogrel, the scientists found. Further research revealed that up to one-third of the general population may have similar variations in this gene, meaning they too probably need a different medicine to reduce heart disease risks.

The findings prompted the U.S.



### Wise Choices Test Your Genes?

Genetic tests aren't necessarily for everyone. You and your doctor might consider analyzing your genes or genome:

- to diagnose a disease.
- if there's a strong family history of a certain disease.
- if you're pregnant or plan to have a child.
- to learn if you have, or are at risk for, an inherited disorder.
- to learn if you're at risk for certain disorders that might be slowed or prevented if you make healthy changes.
- to guide your health care provider in choosing the best treatments for you.

Food and Drug Administration (FDA) to change the label for this common drug to alert doctors that it may not be appropriate for patients who have certain gene variations. Two alternative drugs have since been developed. "If people have these gene variants, they know they have options," says Shuldiner. "This is a great example of how study results made it onto a drug label and are beginning to be implemented into patient care."

Getting a genetic test usually isn't difficult. Doctors generally take a sample of body fluid or tissue, such as blood, saliva or skin, and send it to a lab. Most genetic tests used today analyze just one or a few genes, often to help diagnose disease. Newborns, for example, are routinely screened for several genetic disorders by taking a few drops of blood from their heels. When life-threatening conditions are caught early, infants can be treated right away to prevent problems.

The decision about whether to get a particular genetic test can be complicated. Genetic tests are now available for about 2,500 diseases, and that number keeps growing. Your doctor might advise you to get tested for specific genetic diseases if they tend to run in your family or if you have certain symptoms.

"While there are many genetic



### Definitions

#### Genome

The entire set of genetic instructions in your body.



### Web Links

For more information about personalized medicine, click the "Links" tab at:

<http://newsinhealth.nih.gov/issue/Dec2013/Feature1>

tests, they vary as to how well they predict risk," says Dr. Lawrence Brody, a genetic testing expert at NIH.

For some diseases, such as sickle cell anemia or cystic fibrosis, inheriting 2 copies of abnormal genes means a person will get that disease. But for other diseases and conditions, the picture is more complex. For type 2 diabetes, testing positive for some specific gene variants may help predict risk, but no better than other factors—such as obesity, high blood pressure and having a close relative with the disease.

The latest approach to personalized medicine is to get your whole **genome** sequenced. That's still expensive, but the cost has dropped dramatically over the past decade and will likely continue to fall. Since your genome essentially stays the same over time, this information might one day become part of your medical record, so doctors could consult it as needed.

You can start to get a sense of your genetic risks by putting together your family's health history. A free online tool called "My Family Health Portrait" from the U.S. Surgeon General can help you and your doctor spot early warning signs of conditions that run in your family. To get started, visit <https://familyhistory.hhs.gov/fhh-web/home.action>.

But personalized medicine isn't just about genes. You can learn a lot about your health risks by taking a close look at your current health and habits. Smoking, a poor diet, and lack of exercise can raise your risks for life-threatening health problems, such as

heart disease and cancer. Talk to your health care provider about the steps you can take to understand and reduce your unique health risks. ■

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### National Institutes of Health

Office of Communications  
& Public Liaison  
Building 31, Room 5B64  
Bethesda, MD 20892-2094  
[nihnewsinhealth@od.nih.gov](mailto:.nihnewsinhealth@od.nih.gov)  
Tel: 301-435-7489

**Managing Editor** Harrison Wein, Ph.D.

**Editor** Vicki Contie

**Contributors** Vicki Contie, Alan Defibaugh (illustrations), Dana Steinberg, Carol Torgan, and Katherine Wendelsdorf

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# A Burning Issue

## Handling Household Burns

Accidental burns can happen just about anywhere in your home, and they're not always caused by fire. You might get burned by spilling coffee in your lap, touching a hot iron, or misusing certain cleaning products.

Burns are skin or tissue damage, usually caused by heat. Burns can be caused by hot objects or liquid, fire, friction, the sun, electricity, or certain chemicals.

Each year, about a half-million people nationwide seek medical attention for burns. Household burns lead to nearly 7 of 10 admissions to burn centers. The good news is that the number of deaths from severe burns has dropped by more than half over the past 4 decades, in large part because of treatments developed through NIH-funded research.



### Wise Choices First Aid for Burns

For minor burns:

- Immerse in fresh, cool water, or apply cool compresses for 10-15 minutes.
- Dry the area with a clean cloth. Cover with sterile gauze or a non-adhesive bandage.
- Don't apply ointments or butter; these may cause infection.
- Don't break blisters.
- Over-the-counter pain medications may help reduce inflammation and pain.

Call emergency services (911) if:

- burns cover a large area of the body.
- burns affect the entire thickness of skin.
- the victim is an infant or elderly.
- the burn was caused by electricity, which can lead to "invisible" burns.

The severity of a burn depends on the area it covers and how deep the damage goes. First-degree burns affect only the thin top layer of skin. Second-degree burns include the thick lower layer of skin. A third-degree burn is the most serious; it penetrates the entire thickness of the skin, permanently destroying it and the tissue that's underneath.

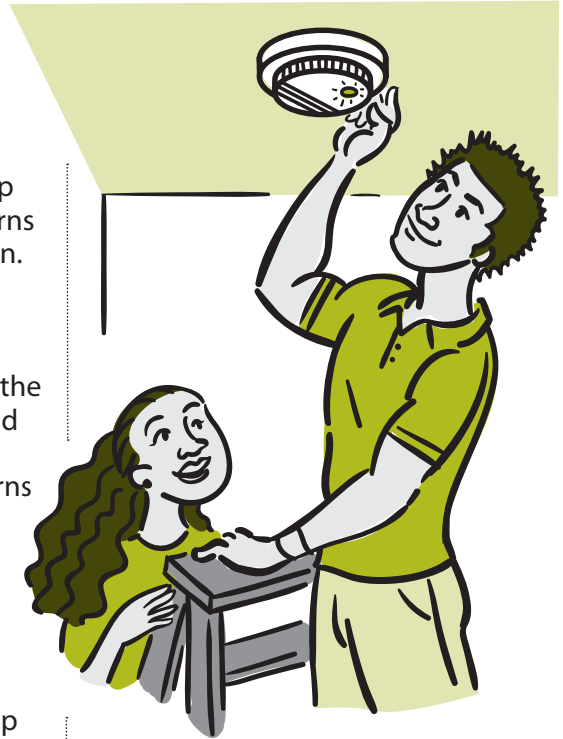
You can care for most minor burns at home. If the burn is red and painful with mild swelling or little blistering, then it's a first-degree or minor second-degree burn.

See a doctor if the burn is dark red and looks glossy with a lot of blistering. These are signs of a deep second-degree burn. Get immediate treatment if the burned skin is dry and leathery, perhaps with white, brown, or black patches. These are signs of third-degree burn.

Burns can become infected with bacteria or other germs if protective layers of skin are lost. Burns can also lead to painful **inflammation**, as your immune system shifts into gear.

"The immune system response is intended to limit the area of injury and to remove any bacteria," says Dr. Ronald G. Tompkins, chief of the burn unit at Massachusetts General Hospital. "But sometimes this immune reaction can lead to further harm to the area damaged by the heat." Proper burn care can help avoid additional damage.

Emergency treatment for third-degree and some second-degree burns may include a blood transfu-



sion and/or extra fluids to help maintain blood pressure. Grafting—placing healthy skin on top of the burn wound—might help promote new skin growth.

Severe burns can lead to widespread inflammation, organ failure, and **shock**. This sometimes-deadly response can arise a week or two after the initial burn. But doctors can't tell beforehand which patients might develop this extreme reaction. Tompkins and other NIH-funded scientists are looking for ways to predict and prevent shock and organ failure after burns or trauma.

You can take steps to avoid household burns. Never leave cooking food unattended on the stove. Set your water heater's thermostat to 120 °F or lower to prevent scalding burns. And install smoke alarms on every floor of your home. Keep yourself and your family safe from unexpected burn injuries. ■



### Definitions

#### Inflammation

Swelling and redness caused by the body's protective response to injury.

#### Shock

A life-threatening condition that occurs when the body doesn't have enough blood flow.



### Web Links

For more about handling household burns, click the "Links" tab at: <http://newsinhealth.nih.gov/issue/Dec2013/Feature2>

# Health Capsules

For links to more information, see these stories online:  
<http://newsinhealth.nih.gov/issue/Dec2013/Capsule1>

## Naps Can Aid Learning in Preschoolers

Afternoon naps can enhance memory and support learning in preschoolers, a new study reports. The finding hints that making time for naps in the classroom might be helpful during early childhood.

Although plenty of studies have shown that overnight sleep and brief naps can boost learning and memory in adults, the effects of napping on toddlers hadn't been closely examined. To learn more, NIH-funded scientists played a memory game with 40 preschoolers. In the morning, the children learned where 9 or 12 cartoon images were located on a grid. Then the children either took an

afternoon nap (about 1 hour and 15 minutes) or were gently kept awake.

After nap time, the children were tested to see how well they could remember the locations of the cartoon images. The scientists found that children could recall 10% more of the items' locations when they napped than when they'd been kept awake. Children who'd napped had similar success in remembering items' locations even the next morning. The researchers also found that the benefits of napping were greatest for the children who regularly took naps.

To explore how memories might be stored in the brain, the team mea-

sured the brain waves of 14 additional children during naps. The researchers noticed a link between distinct bursts of brain activity during napping and a child's performance on memory tests. These bursts of activity might represent the strengthening of memories, the scientists suggest.

"We hope these results will be used by policy makers and center directors to make educated decisions regarding nap opportunities in the classrooms," says the study's lead researcher, Dr. Rebecca Spencer at the University of Massachusetts Amherst. ■

## Resource for People with Down Syndrome

NIH has launched a free, confidential, Web-based health registry for people with Down syndrome and their families and caregivers.

The registry, called DS-Connect, offers a secure place for people with Down syndrome—or family members on their behalf—to enter and save basic health information. It also can help families stay informed about potential treatments and other efforts to improve the quality of life for people living with Down syndrome.

Down syndrome is one of the most common genetic birth defects nationwide. People who have Down syndrome have a characteristic facial appearance and intellectual difficulties. They may also face other health issues, such as digestive problems,

heart defects, and hearing loss.

The basic medical information that participants provide on DS-Connect can be studied by scientists to help them learn more about Down syndrome. The website's design ensures that this information remains anonymous and confidential.

If participants give permission to be contacted, the registry coordinator can let them know about clinical research studies they might qualify for. Such studies might eventually lead to improved treatments or better quality of life for people with Down syndrome.

To learn more about DS-Connect, or to create a personalized profile for your loved one with Down syndrome, go to <http://DSConnect.nih.gov>. ■



### Featured Website NIH Health Information

<http://health.nih.gov/>

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