WHAT ARE THE SYMPTOMS OF FABRY DISEASE?

Someone with Fabry disease may have only one symptom or a mix of several symptoms. Symptoms may range from mild to severe.

One of the most common symptoms of Fabry disease is pain, tingling, or burning in the hands and feet, typically beginning in childhood. Other symptoms include:

- Feeling tired or weak
- Not sweating enough
- Red-purple skin lesions (spots on the skin), most often seen in the bathing trunk area
- Abnormal pattern of lines on your cornea (the outer layer of the eye) seen by your eye doctor during an eye exam
- Frequent fevers
- Being sensitive to hot and cold temperatures
- Stomach problems (abdominal cramping, pain after eating, diarrhea, constipation, nausea, vomiting)
- Heart problems (low or high blood pressure, irregular heartbeat, heart failure, enlarged left heart)
- Stroke or transient ischemic attack (TIA)
- Kidney problems (protein in the urine, kidney disease)
- Nervous system problems (vertigo, feeling weak, dizzy or numb, hearing loss, ringing in ears)
- Depression and anxiety

Because the disease is linked to an X chromosome, females may show no symptoms, which is why testing is so important if you suspect that you may have Fabry disease.

CAN PEOPLE WITH FABRY DISEASE BE TREATED?

If you have Fabry disease, a team of specialists can help you manage your symptoms. Your care team may include a:

- Cardiologist (heart doctor)
- Neurologist (nervous system doctor)
- Nephrologist (kidney doctor)
- Gastroenterologist (digestive system doctor)
- Audiologist (ear doctor) or otolaryngologist/ENT (ear, nose and throat doctor)
- Psychologist, psychiatrist or mental health counselor

There are guidelines that have established what conditions and markers should cause a person to start treatment. Talk to your doctor about a disease management plan that is best for you.

IS THERE A TREATMENT FOR PEOPLE WITH FABRY DISEASE?

If you have Fabry disease, treatment options are available. Talk to your doctor about a disease management plan that is best for you.

RESOURCES:

DiscoverFabry.com

KidneyFund.org/fabry

American Kidney Fund®
FIGHTING ON ALL FRONTS

sanofi

MAT-US-2302786

Talk to your doctor about

FABRY DISEASE

Diabetes and hypertension are not the only causes of kidney disease;
IT COULD BE SOMETHING ELSE.

There are many genetic causes of kidney disease. One of them is a disorder called Fabry disease.

FABRY AND YOUR KIDNEYS

Intended for US residents only

American Kidney Fund®
FIGHTING ON ALL FRONTS

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WHAT IS FABRY DISEASE?

Fabry disease is a rare genetic disorder that causes a fatty substance called GL-3 to build up in certain parts of the body, like the kidneys, heart, and skin. When you have Fabry disease, your body does not make enough of an enzyme called alpha-galactosidase A (alpha-GAL). Your body needs alpha-GAL to help break down a fatty substance in your cells called globotriaosylceramide (GL-3).

Fabry is a genetic disorder that can be passed down from parent to child. Several members of the same family often have it. Genetic disorders are caused by a change in one of your genes. The gene associated with Fabry disease is called *GLA*.

HOW DOES FABRY DISEASE AFFECT YOUR KIDNEYS?

Fabry disease causes GL-3 buildup in some cell types, causing tissue damage in certain organs; including the kidney. The GL-3 buildup in the kidneys leads to kidney damage and gets in the way of the kidneys' ability to filter waste from the body.

Kidney damage cannot be reversed and worsens over time. This is why early diagnosis is so important. The earlier you find out you have Fabry disease, the sooner you can start tracking your symptoms and learning how to manage them.

TALK TO YOUR DOCTOR

If you think you or someone in your family may have Fabry disease, talk to your primary care provider or nephrologist. Even if you do not have additional Fabry symptoms but have unexplained chronic kidney disease (CKD) or kidney failure, consider asking about genetic testing for Fabry disease.

Here are some ways to start a conversation about Fabry disease testing at your next appointment:

1. Someone in my family has kidney disease or Fabry disease. Should I get tested for Fabry disease?
2. I'm having symptoms that I think could be Fabry disease. Should I get tested?
3. I never got a reason for why my kidneys failed. Could it be Fabry disease?

Not all doctors are experts on Fabry disease. Your doctor may suggest you talk to another doctor or specialist to get your questions answered.

SHOULD YOU GET TESTED FOR FABRY DISEASE?

When 1 person is diagnosed with Fabry disease, an average of 5 other family members may be affected as well. This can include brothers, sisters, children, parents, aunts, uncles, and cousins. Remember, both males and females are at risk. You should consider getting tested for Fabry disease if:

- You do not know the cause of your chronic kidney disease (CKD) or end-stage renal disease (ESRD/ kidney failure)
- You think you have symptoms of Fabry disease
- Someone in your family has Fabry disease or thinks they have symptoms
- Someone in your family has CKD or kidney failure

Even if you have never heard of Fabry disease, you could still be at-risk and should consider getting tested.

HOW DO DOCTORS TEST FOR FABRY?

Doctors can test for Fabry disease with a blood or saliva test where testing for Fabry disease differs by sex. Talk to you doctor about which test you will need.