Fabry disease is an X-linked disorder. It is caused by a mutation in a gene on the X chromosome and can be passed down by either parent.

Why do mutations matter?

The type of mutation may help predict when symptoms appear, what kind of symptoms appear, and how bad the symptoms are or may become.

It’s important for individuals or families with Fabry to know which mutation they have.

At least 1000 different gene mutations have been identified that can cause Fabry disease.

HOW FATHERS PASS ALONG FABRY DISEASE

Men have one X chromosome and one Y chromosome. Women have two X chromosomes. A father with Fabry disease passes his mutation to all of his daughters, because daughters inherit their father’s only X chromosome. An affected father never passes the mutation to his sons, because sons inherit a Y chromosome from their fathers.

HOW MOTHERS PASS ALONG FABRY DISEASE

A mother who has the mutation on one of her two X chromosomes has a 50% chance of passing down Fabry disease to each of her children. Since men only have one X chromosome, if they inherit the mutation, they will develop Fabry disease. In affected daughters, the α-Gal A mutation can occur randomly in some cells and not others, so daughters may have a broader range of variable symptoms than sons.
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In Fabry disease, the affected enzyme is α-galactosidase A, or just α-Gal A. Fabry is a rare, progressive, genetic disease affecting 1 in 117,000 people, although it may be more common than this.

It's a type of disease called a lysosomal storage disorder, or LSD. People with LSDs have problems making specific lysosomal enzymes.

Usually, α-Gal A breaks down substances in the cell called globotriaosylceramide (GL-3) and plasma globotriaosylsphingosine (lyso-Gb 3). In people with Fabry disease, α-Gal A does not break down GL-3; instead, GL-3 builds up, particularly in cells lining blood vessels. This buildup damages tissues and organs, leading to the symptoms of Fabry disease.

Breaking down Fabry: facts about the disease

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Sometimes, mutations occur in the DNA code that makes up a particular gene.

Think of it like spelling. One wrong letter can completely change the meaning of a word!

Every person has information coded into his or her cells that's called DNA—which is inherited from his or her parents.

On a molecular level, everyone is unique. Every patient is unique.

For additional information, talk to your healthcare provider.

A doctor suspects Fabry disease—here's an example of what can happen

How does Fabry disease affect the body?

From one person to another, everyone experiences Fabry disease differently.

Psychosocial
- Feelings of guilt, apprehension, depression, isolation, and even fear (may also be experienced by family members)
- May experience feelings of relief to finally have a diagnosis of Fabry, after years of not knowing
- Shock may also be felt from receiving a diagnosis of Fabry

Nervous system
- Pain
- Hearing loss, ringing in the ears
- Intolerance to heat, cold, or exercise
- Transient ischemic attack (TIA) and stroke
- Burning in the hands and feet, also called acroparesthesia
- Vertigo/feeling dizzy

Heart
- Irregular heartbeat (fast or slow)
- Heart attack or heart failure
- Enlarged heart

Gastrointestinal
- Nausea, vomiting, cramping, and diarrhea
- Pain/bloating after eating, feeling full after a small amount of food
- Constipation
- Difficulty managing weight

Skin
- Sweating less than or more than normal
- Small dark red spots called angiokeratomas, particularly between belly button and knees

Kidneys
- Protein in the urine
- Decreased kidney function
- Kidney failure

Eyes
- A whirled pattern in the cornea
- Fabry cataracts

Bottom line: Talk to others with Fabry disease and share your experiences. It may help!

What causes Fabry disease?

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What causes Fabry disease?
What do these words mean? How does Fabry disease affect families?

Fabry disease is an X-linked disorder. It is caused by a mutation in a gene on the X chromosome and can be passed down by either parent.

A glossary of important terms when discussing Fabry disease:

- **Cell**: Basic building block of all living things
- **Chromosomes**: Structures that contain DNA and a person's genetic code
- **De novo mutation**: An alteration in a gene that is not inherited but is present for the first time
- **Deoxyribonucleic acid (DNA)**: Basic unit that allows for the transmission of genetic information from one generation to the next and contains instructions, or code, for making proteins and enzymes
- **Enzyme**: A special type of protein that speeds up a reaction that takes place within a cell
- **Lysosome**: A specialized fluid-filled sac, found in cells, that contains enzymes
- **Lysosomal storage disorder (LSD)**: A group of over 50 diseases resulting from the accumulation of waste products in lysosomes
- **Mutation**: A permanent error in the DNA code
- **X-linked disorder**: Inherited disorder caused by a mutation in a gene on the X chromosome

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