

What is Mitochondrial Disease?

Every part of your body is made up of very tiny things called cells. On average there are around 37 trillion cells in a grown person. That's 37,000,000,000,000 cells! You need a microscope to see a cell. In fact, they are so small that thousands of cells would fit inside the period at the end of this sentence. That's tiny!

The cells are designed to do all the different jobs our body needs. There are skin cells, blood cells, hair cells, muscle cells, brain cells, stomach cells... all the different kinds needed for a body to work. Inside most of these cells, there's an oval-shaped piece called the mitochondria. It has several jobs, but the main job is to make energy. Each cell needs energy to do the job it was made for. The mitochondria use the food and oxygen we eat and breathe to make that energy.

Someone with mitochondrial disease has a problem in the mitochondria. It can't make energy the right way. This is not a disease you can catch, like a cold or the flu. It must be inherited from your parents, or their parents, or someone in the family history - the same way you get your eye color, the color of your hair, and how tall you will be.

Because so many different kinds of cells exist, there are many different kinds of mitochondrial disease. Body parts that need the most energy are usually most affected. Those are your brain, nerves, muscles, heart, pancreas, liver, kidneys, eyes and ears. Often several of these body parts will have problems depending on what type of mitochondrial disease the person has. The United Mitochondrial Disease Foundation (UMDF) lists 47 broad types of mito disease on its website, many with the same symptoms. But there are hundreds of variations, just like there are many kinds of cancer. It is estimated that 1 in 5000 people are born with a genetic mitochondrial disease.

My son, upon whom Peter in this story is based, had PolG. It mainly affected his muscles, brain, liver, eyesight and digestion. Peter's symptoms started after he turned four. We worked with doctors to manage his symptoms and not make his body need more energy than it could create.

Like my son, Peter, many people with mito disease take specific vitamins and supplements that can help the body struggle less with making energy. But it does not fix the problem. So far, individuals with mitochondrial disorders have no cure for their disease. The younger a person is when symptoms start, the shorter their life will likely be.

Scientists who study the mitochondria have made steady progress in identifying the diseases caused by mitochondrial dysfunction and in managing the symptoms or effects of the disease to slow down its progress. But more research is needed to find a cure.

Check out these resources for more information about Mitochondrial disease:

United Mitochondrial Disease Foundation-

<https://www.umdf.org/>

Phone: 412-793-8077 or 888-317-8633

MitoAction- <https://www.mitoaction.org/>

Phone: 888-648-6228

Cleveland Clinic-

<https://my.clevelandclinic.org/health/diseases/15612-mitochondrial-diseases>

National Organization for Rare Disorders (NORD)-

<https://rarediseases.org/>

Phone: 203-744-0100

Genetic and Rare Diseases (GARD) Information Center-

<https://rarediseases.info.nih.gov/>