Urea Cycle Disorder Facts

A urea cycle disorder (UCD) is a rare disease that affects about 1 in 35,000 births in the United States. Just because it's rare doesn't mean there isn't information to help you or someone you love who is living with a UCD. Here are some facts to help you better understand a UCD.

IT'S IN THE GENES

A UCD is a genetic disease that is passed on to a child. See below to learn how it happens and what genetic testing can show.



A UCD is something a person is born with.

Everybody has 2 copies of each gene, 1 from the mother and 1 from the father.





UCDs can be passed down when each parent gives the defective gene to the child. One UCD subtype, OTC, can also be inherited in an X-linked way, where the gene is passed on from the mother. UCDs can also be caused by a spontaneous genetic mutation.

The UCD subtype, ornithine transcarbamylase (OTC) deficiency, is the most common and is passed along the X chromosome from the mother, who may not know she is a carrier. Or in rare cases it can be passed on from a father to his daughter.



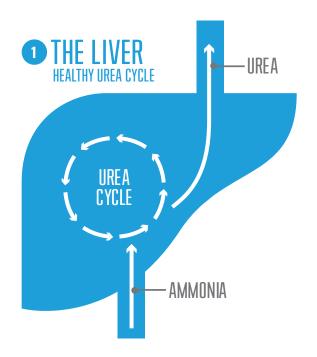
A carrier is a person who has 1 normal copy of the gene and 1 abnormal copy.

Genetic testing can discover if a person is or isn't a carrier and determine the chances of passing the defective gene on.





THE UREA CYCLE AND UCDS



Healthy urea cycle

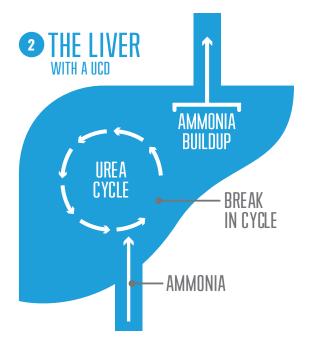
Protein found in food enters the body to be absorbed by cells for growth and development.



As our bodies break down protein into amino acids, leftover nitrogen from the protein is turned into ammonia.



The ammonia is then removed from the blood, turned into urea, and excreted from the body through urine.



Urea cycle disorder

When a person has a UCD, the urea cycle can't convert the ammonia safely into urea and the ammonia builds up because the body can't get rid of it.

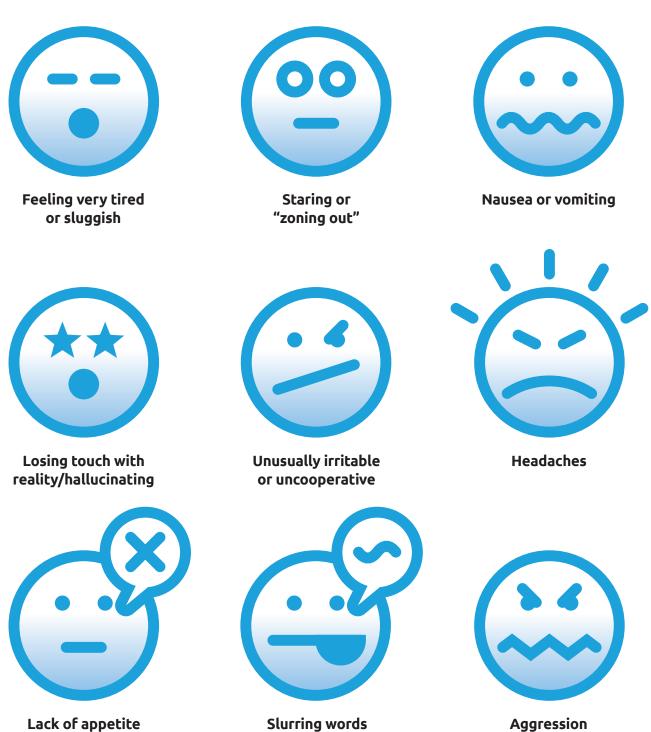


The ammonia can reach toxic levels in the blood and can trigger vomiting, confusion, and swelling in the brain.



SPOT THE SYMPTOMS OF A UCD

Normal things in life, like being sick, stressed out, and eating too much food high in protein, can cause ammonia to rise to toxic levels. This can trigger a hyperammonemic crisis, which requires immediate medical attention. Take a look at these common symptoms of high ammonia levels that could signal a crisis.





MANAGING YOUR UREA CYCLE DISORDER

There are options to help control high levels of ammonia associated with a UCD. Because every patient's needs are different, it's important to talk to your doctor to develop a plan on how you can best manage your ammonia levels.

Low-protein diet

It's important to limit protein when living with a UCD. But limiting it too much may harm your body at the same time because you need it to grow. Common triggers, like being stressed or getting sick, can increase ammonia to toxic levels despite your best efforts to balance protein intake.

Amino acid supplements

Amino acids are essential nutrients that are needed to grow and maintain your health. The body creates amino acids when it breaks down protein. If you are on a low-protein diet, amino acid supplements can fill the gap for the nutrients your body cannot make itself. A dietitian who is part of your healthcare team can help decide which supplements may be right for you.

Nitrogen scavengers

When the urea cycle can't get rid of ammonia, a treatment that gets rid of the excess ammonia may be needed. Nitrogen scavengers, sometimes called "ammonia-removing medicines," help remove ammonia from the bloodstream. Nitrogen scavengers come in many forms, including capsules, powder, and liquid injection.

Liver transplant

Living with a UCD means that certain enzymes in your liver aren't working properly. A liver transplant can cure a UCD so the enzymes work the way they should. The transplant can't fix any health problems that happened beforehand, and the procedure has its risks.



