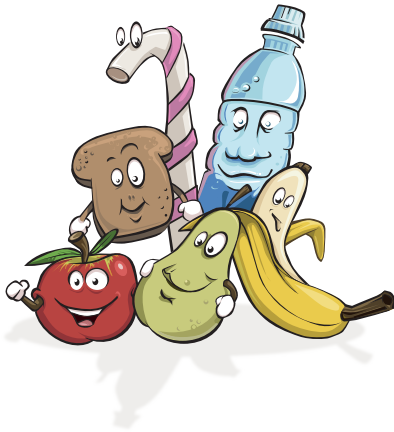




***Urea cycle disorders and organic acidurias
For younger people***

What is a Urea cycle disorder/organic aciduria?

The food we eat gets broken down by the body with the help of thousands of chemical reactions to produce energy and to help us grow and remain healthy. You may remember that the food we eat is made up of carbohydrates, fats and protein. Carbohydrates and fats give us energy, and protein helps us grow, build our bodies and remain healthy.

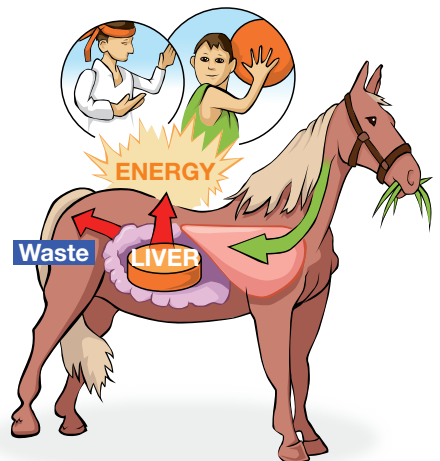


*Protein, Carbohydrates, Fat
(and vitamins, minerals and water)*

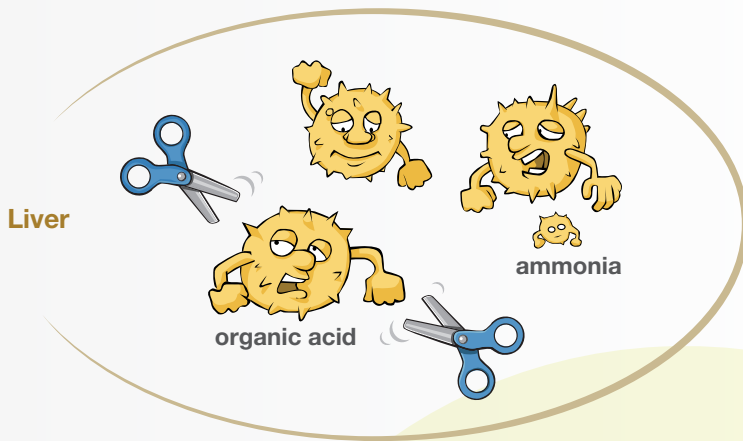
Your body needs all of these

People with a urea cycle disorder or organic aciduria can get sick if they eat too much protein from certain kinds of food (this is why they need a special diet).

Some examples of food that give us protein are milk, eggs, meat, fish, beans and nuts. Protein from the food we eat gets broken down and sent to the liver to be sorted. The liver decides how much is needed by the body and how much should be got rid of as waste.



Your liver also takes toxins (harmful substances) out of your body. Excess protein that the body does not need is broken down to produce two harmful substances, called “ammonia” and “organic acids”. The liver quickly gets rid of these with the help of chemical reactions. The chemical reactions are carried out with the help of special proteins called enzymes, which are like scissors – they help “cut” chemicals to form other chemicals.



- ammonia is converted to harmless substance, urea which is passed out in the urine (that's why it is called urine!).
- organic acids are recycled to make energy or other proteins which can be used by the body.

When something goes wrong in these chemical reactions, there is a block in the system and ammonia or organic acids build-up and overflow from the liver into the rest of the body, particularly the brain, where they can cause harm and make you feel ill.

You could imagine that your bathtub is your liver and that the water is ammonia or organic acids – if you fill up the bathtub with water, and keep filling it but don't take the plug out, it will overflow...



This is what happens in a urea cycle defect or organic aciduria. Your special diet and medicines will help you get rid of the excess ammonia and organic acids to keep you healthy.

Why has it got a funny name?

Depending on exactly where the chemical reactions are blocked, the urea cycle defects and organic acidurias are given special names. Doctors like to make things sound more complicated than they really are!



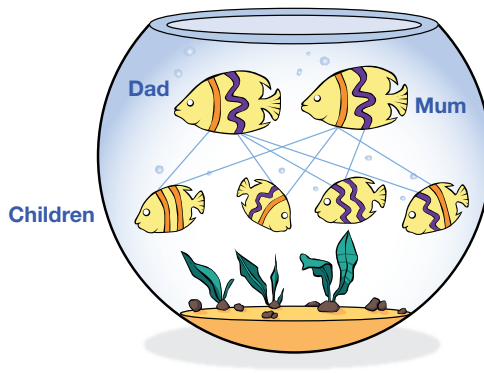
These names simply tell us which reaction is blocked or which chemicals overflow from the liver into the body and urine. It really does not make a big difference to you or your treatment!

How did I get this disease?


It is nobody's fault. A Urea Cycle Defect/Organic Aciduria is what we call a genetic disease; this means something you are born with.


Information on genes and genetic diseases


When a child is conceived, one egg from mum and one sperm from dad is brought together to form the baby. There is no way of knowing which sperm and which egg will unite to make the baby. Mum's egg and dad's sperm carry information ("genes") that is responsible for the colour of your hair, eyes, etc. and will also carry information which may be related to genetic disease.



 Normal gene
  Gene with disorder

 Does not have a disorder

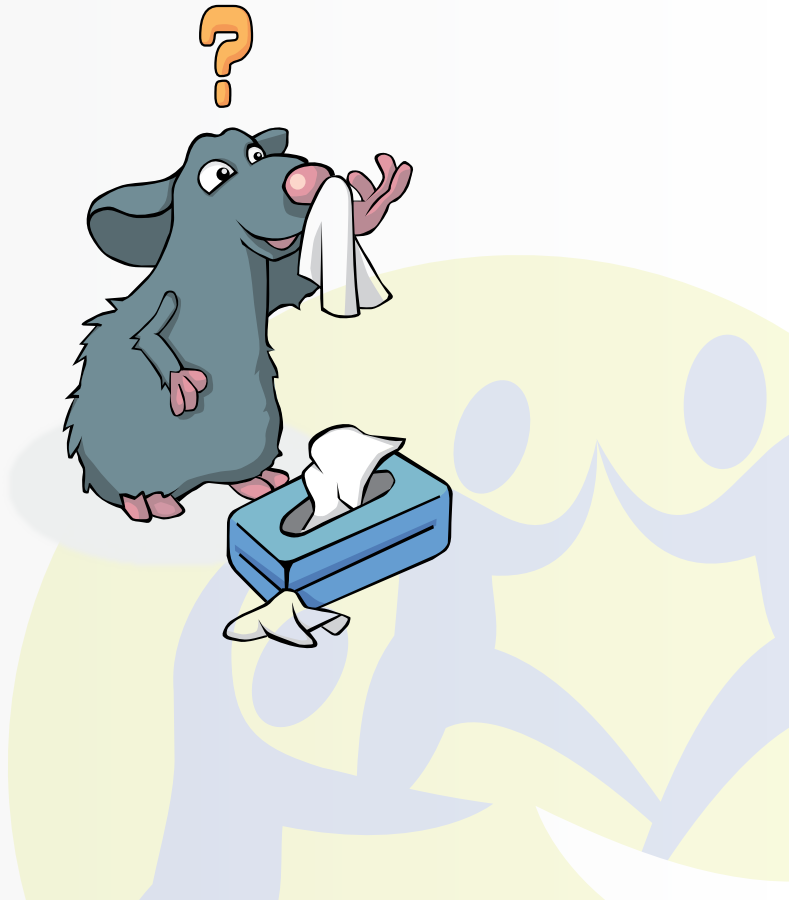
 Does not have a disorder but carries one defective gene like mum and dad

 Carries 2 defective genes and has a disorder

Most UCDs and OAs arise when mum's egg and dad's sperm both "carry" information about a genetic fault which is passed on to the baby.

Is it catching?

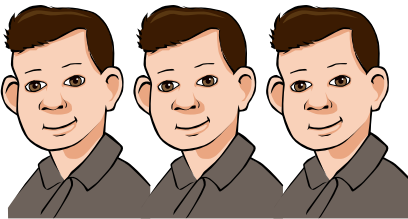
No, it is not catching. It is not due to a virus or a bug; it is due to a mistake in the genes.



Am I different from everybody else?



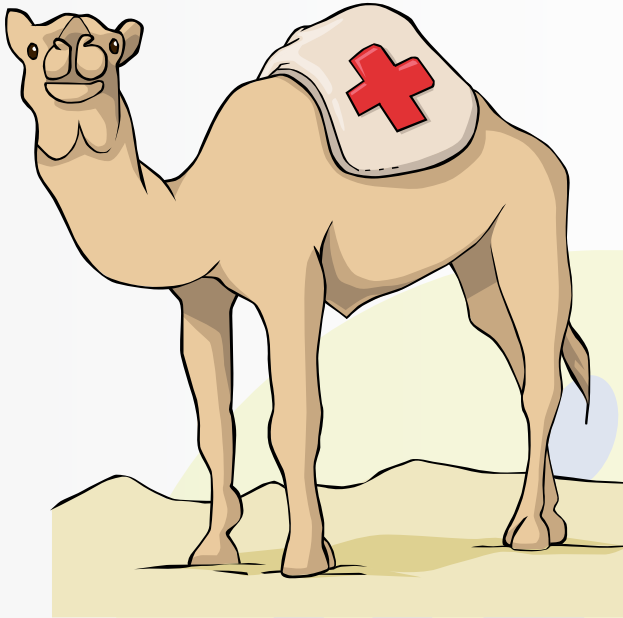
Everyone is different. Some of us are tall and some short, some clever at school work, some clever at other things. A person with a UCD/OA just has a medical problem needing treatment.



Imagine if we were all the same!

Why do I have to be on a special diet? Why do I have to keep taking medicines?

The special diet controls the amount of harmful substances in the body and keeps you well. It may be hard to stick to the special diet and keep taking the medicines, but not taking them will make you feel sick and unwell.



How long do I have to keep taking the diet and medicines?

You will have to keep taking the diet and medicines forever. This sounds like bad news, but this is better than feeling sick and unwell, isn't it?



What will happen if I take my diet and medicine everyday?

You will feel much better and you will be able to do much more and have much more fun!

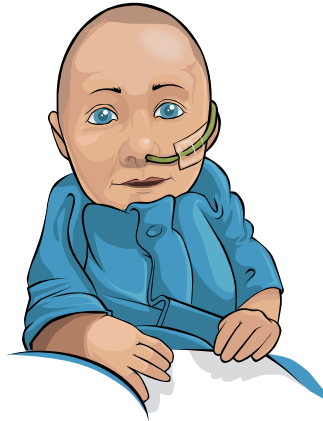


What will happen if I stop taking my diet and medicines and eat things I am not supposed to?

The levels of ammonia and organic acids in your body will build up and you may feel very ill indeed! You may need to come into hospital for special medicines. Do not stop taking your medicine and stick to your diet!

Why do some people with UCD/OA need a feeding tube?

People with UCD and OA often do not feel like eating, and the amount they eat may not be enough to stay healthy and grow well. The tube can also be helpful if you do not like the taste of the medicines. The tube is also very helpful in giving food and medicines when you are feeling sick and unwell.



A feeding tube supplies food or medicine directly to the stomach.

The use of a feeding tube is different for every person and you should discuss this with your metabolic team.

Will my problem get better or worse?

The problem is due to a mistake in the genes, so it remains throughout life. Medicines and your special diet help you deal with it so you feel well. There may be some times when the body produces more ammonia or organic acids than usual – this usually happens when you have another illness, such as a cold, fever or a sickness bug. This happens for a short time and you may feel unwell during this time.



What do I do?

When you have a cold, fever or sickness bug you will require a special diet called “emergency regime” to help you feel better quickly. Sometimes you may feel very ill and need to come to hospital for a few days for special medicines to get you better.

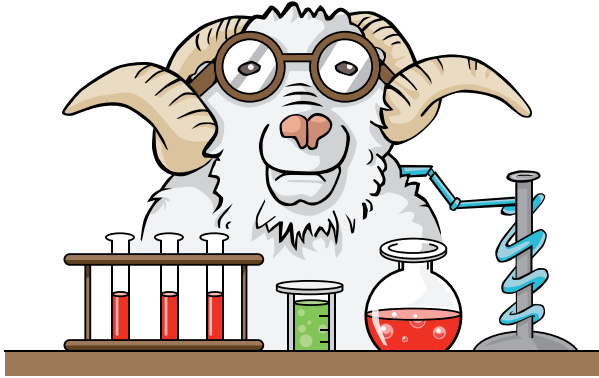
Will it hurt?

No, it will not hurt but you may feel sick and unwell.

Will this happen every time I fall ill?

Everybody is different, every illness is different and the way the body deals with illness is different every time. Some illnesses may make you feel sicker than others, whereas some may not make you feel ill at all. As it is impossible to tell, you should take any illness seriously and use the “emergency regimen”.

Why does my doctor keep doing blood tests?



Blood tests are needed to check how well your body is working and whether the treatment is working.

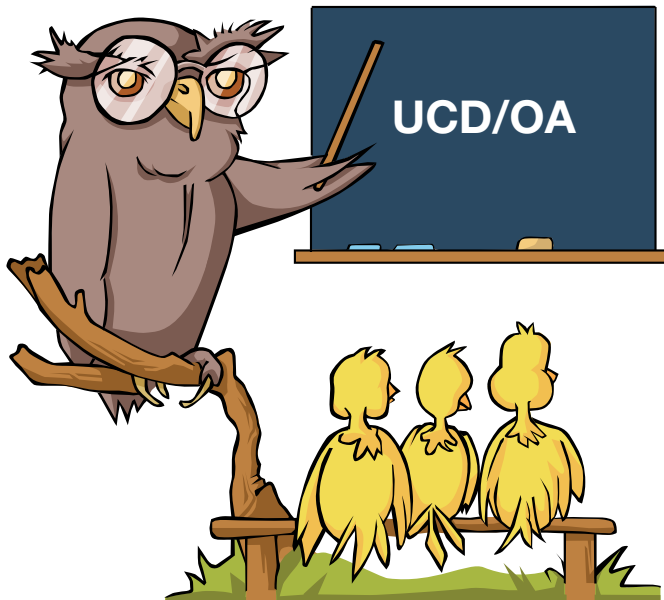
Am I allowed to.....?

- play on a computer? **YES**
- play games? **YES**
- go to parties? **YES**
- go on holiday? **YES**
- Go to college? **YES**



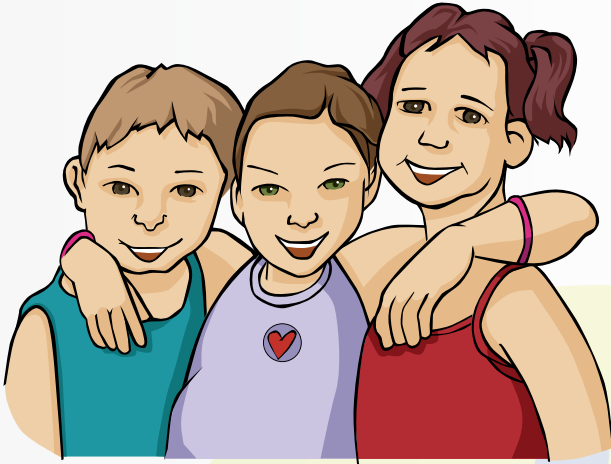
Do other people need to know?

It is probably best that your schoolteachers know that you have this problem and your parents will probably talk to them about this.



Do my friends need to know?

It is up to you to whether or not to tell your friends. It is usually best to tell your good friends that you've got a medical problem, so they understand why you need a special diet and medicines.



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For more information: http://ec.europa.eu/health/programme/policy/index_en.htm

E-IMD would like to thank the Eurowilson network for kindly sharing the illustrations used in this booklet www.eurowilson.org.

For more information and contacts of patient organizations: www.e-imd.org