

NEPHROPATHIC

CYSTINOSIS



*explained
to children*

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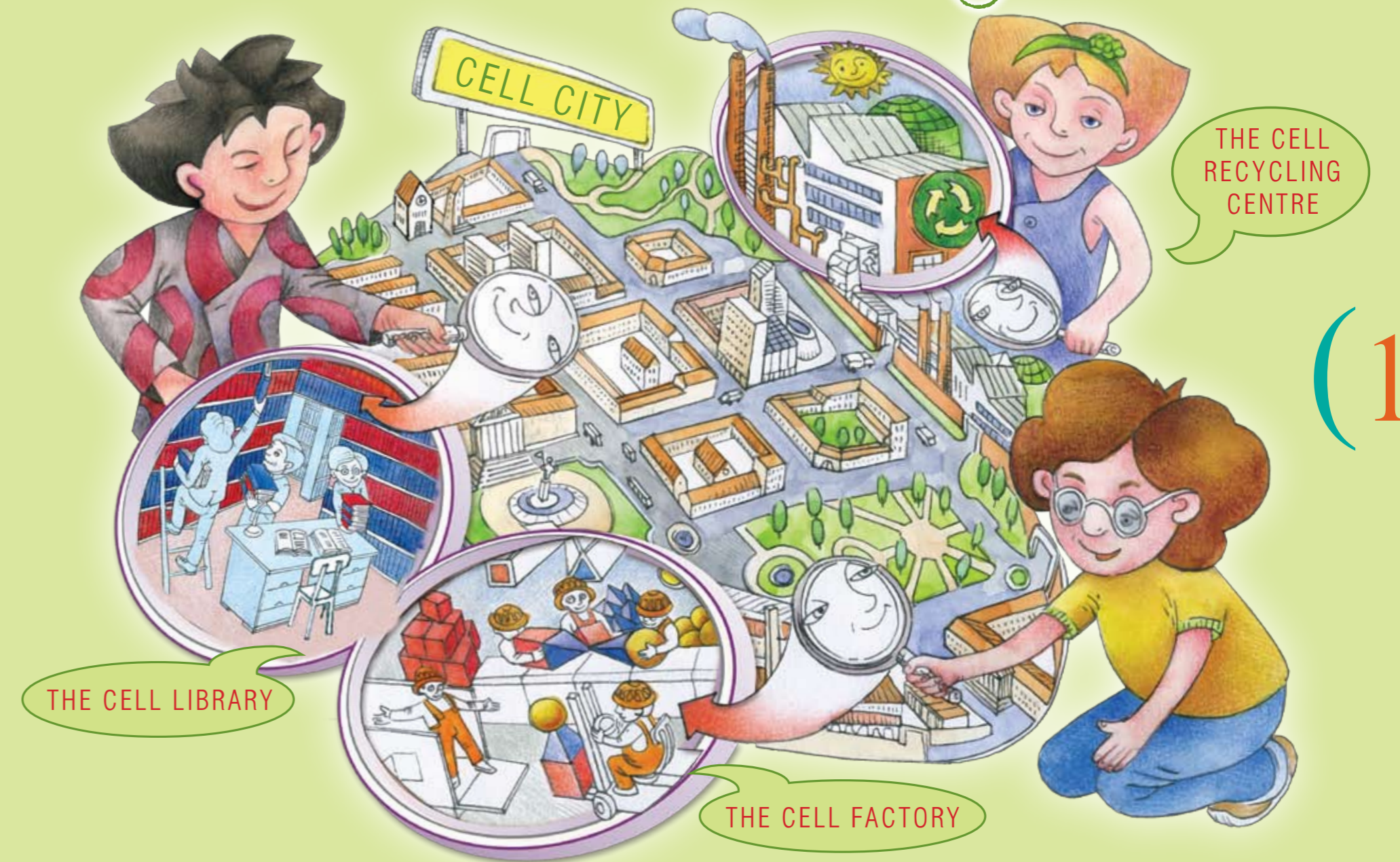
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An introduction to cystinosis



1. The human body

Your body is a marvellous thing! It is made of lots of different parts like skin, bones, muscles, eyes and ears. Each of these parts is made up of lots and lots of cells. Cells are so very small that we cannot see them unless we use a very strong magnifying glass.



Each cell has special jobs to do. For example, there are cells in your eyes that help you see and cells on your tongue that let you taste sweet things like sugar or sour things like lemon juice. Sometimes some of these cells do not work properly. So, if some cells in your tongue cannot work well, you would not be able to taste the sweetness of sugar or candy.

In children with cystinosis, some of the body's cells do not work properly and in this book we are going to explain what happens. So to understand what cystinosis is, we need to look at how the cells of the body work.

Let us take a close look at the inside of a cell. Imagine the cell as a factory with lots of different parts making and sorting different things.

At the centre is the cell library (the "core") which is where all the instructions for how the cell works are kept.

There are books that contain maps for transport trucks that carry things around in the cells and instructions for making new building blocks for the body cells. There are even instructions for the colour of your hair!

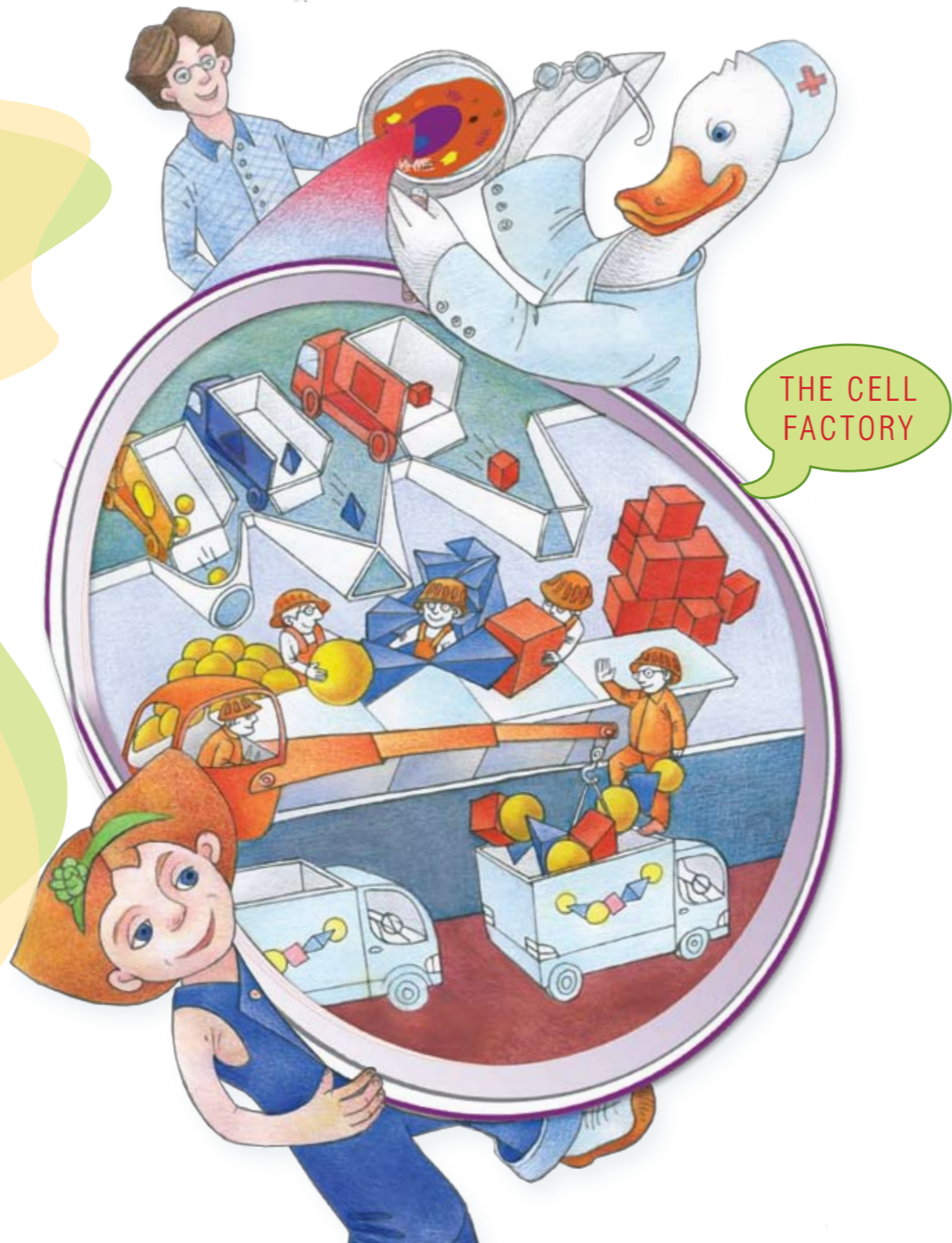
THE CELL LIBRARY



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In another part of your cells there is an assembly line where the building blocks can be made and sent around the cell to make new parts. In this way the cells can make new cells or parts of them. This is done to help you grow but also to repair old or damaged cells. This way your body can stay fit and healthy. .

Of course every good factory has to get rid of its rubbish or waste. Just like in houses where some rubbish is taken away in sacks and cans, glass and paper can be sent to recycling centres, the body cells have ways of dealing with waste. Anything in your cell that is old or not needed any more is taken to a special place, where it is broken down and most of the building blocks can be re-used. To do this, every cell has its very own recycling centre. The cell has ways of taking the waste to the recycling centre, where it is taken apart and sorted into parts that can be re-used and others that need to be thrown out.



2. What cells are made of

The walls that surround cells and many of the special parts inside are made of proteins. Proteins are everywhere! Meat, fish, milk, bread and lots of other foods have lots of protein inside the cells.

The food we eat is broken down in our tummies so the proteins can be used by our body. Our bodies use the food protein to help build new cells (that is what helps us to grow up).



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In order for your body to use the proteins from the food you eat, proteins have to be broken down even further into the tiny little blocks that they are made of. These parts of proteins are called “amino acids”, they come in different shapes and sizes and one of these amino acids is called cystine. You will find cystine in lots of different proteins, in food and in our cells.

3. How does the cells recycling center work?

As well as using protein from food to build cells, the body can re-use the amino acid building blocks from proteins in old cells to make new cells – think of how we recycle plastic, cans and paper. So as we showed you on the picture of the cell earlier, the body has built-in recycling machines. This building of new proteins and re-cycling of old ones goes on in cells

all the time without you even thinking about it. So the cystine in our body comes partly from food we eat and partly from the body recycling old proteins.

Specialised transport trucks bring the proteins the cells do not need any more to the recycling centre. Once they are unloaded, the proteins are being broken up into their smaller building blocks - the amino acids. The breaking down of proteins needs powerful chemicals that need to be kept secure in the recycling centre – like keeping dangerous cleaning fluids locked in a cupboard. Just as in any recycling centre, the amino acids are then sorted. Many amino acids are still good and can be sent to the cell’s assembly factory to build up new proteins.



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In order to get there, the amino acids need to get out of the recycling centre. They leave it through very special doors which will keep the dangerous chemicals inside but let out the safe amino acids. The shape of each door makes sure that only one kind of amino acid can get through. Cystine, for example, can only get out through the door that is shaped like cystine.

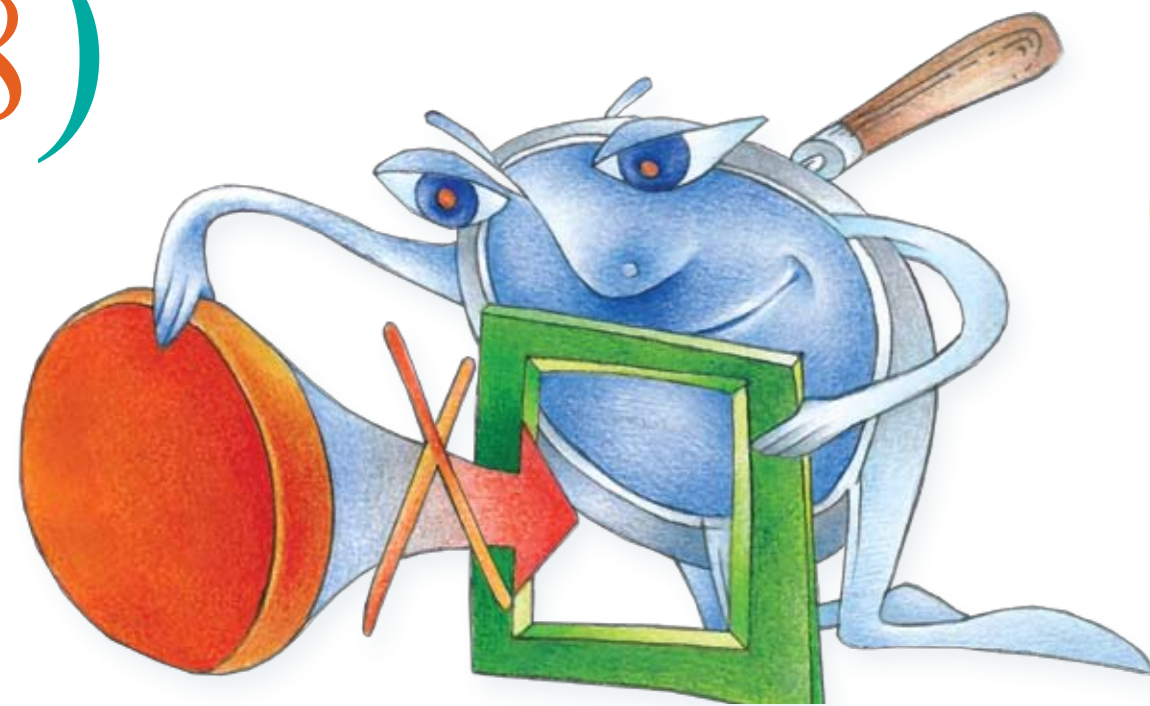
Just like a round peg does not fit a square hole, cystine can only leave the recycling centre through the cystine door.

It took a long time to find out the problem in cystinosis. Now we know that the recycling centres in cystinosis cells have a problem getting the cystine out, as if the cystine door cannot open. Imagine if you could not recycle or throw away your waste at home or at school – it would cause quite a mess!

If the recycling centres' door for cystine does not work, more and more cystine is left inside and starts taking up more and more space. At some point, it will take up so much space, that the recycling centre cannot work properly anymore.

And if the recycling centre does not work, the whole cell will slow down – like a factory full of waste, it cannot work properly. If many cells stop working properly, the part of the body made of these cells will not be able to function so well.

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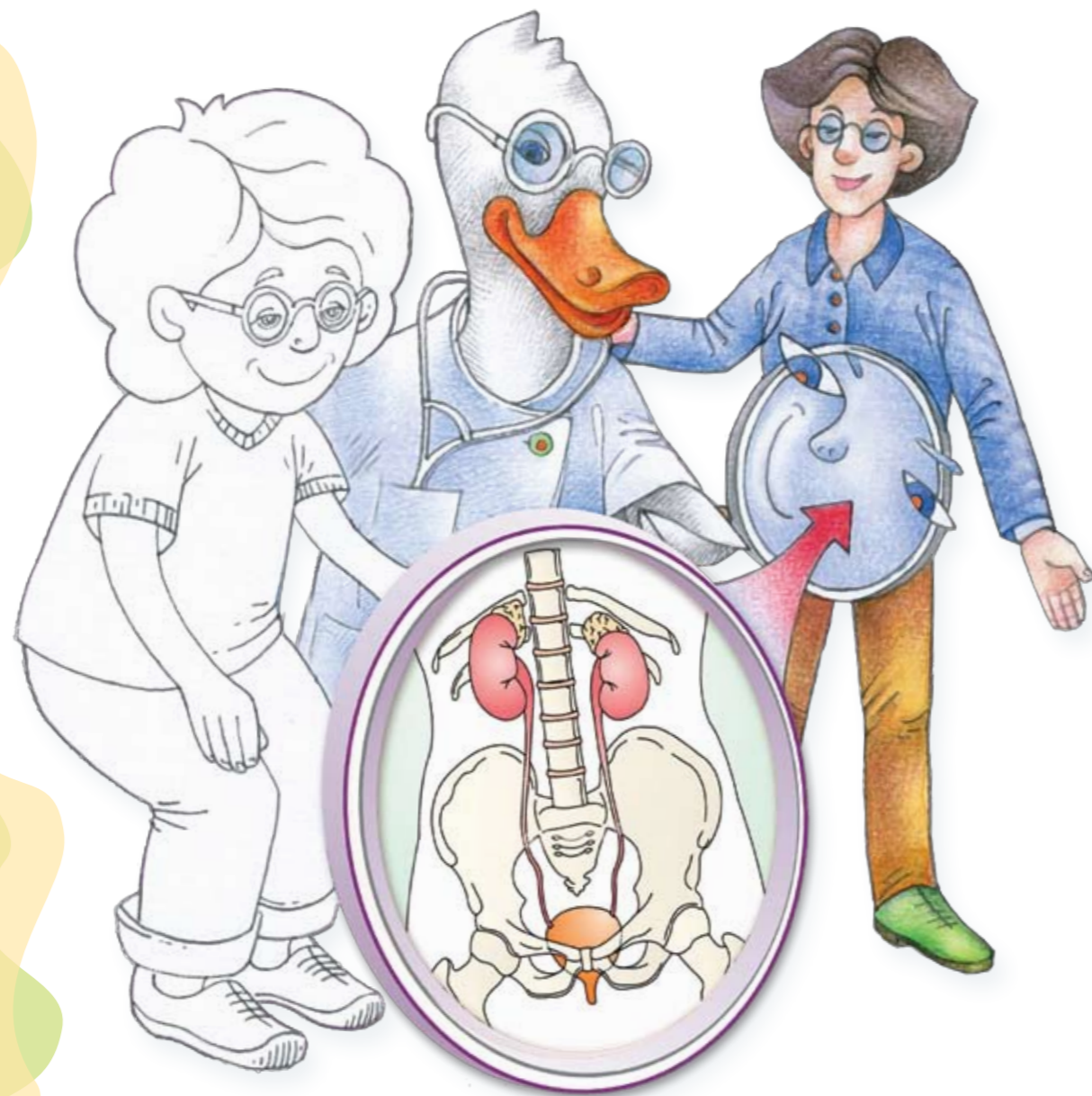
THE CELL
RECYCLING
CENTRE



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Although recycling centres are present in every cell in the body not all cells will be damaged in cystinosis. When cystine builds up, there are parts of your body that get hurt more than others – like the kidney and parts of your eyes.



The kidneys get damaged in cystinosis because the cells in the kidneys have many recycling centres. Children with cystinosis often first know that they have cystinosis because they have problems with their kidneys. But even though these cells do not work so well and can make you feel poorly, lots of other body cells are working really well.

The kidneys and cystinosis



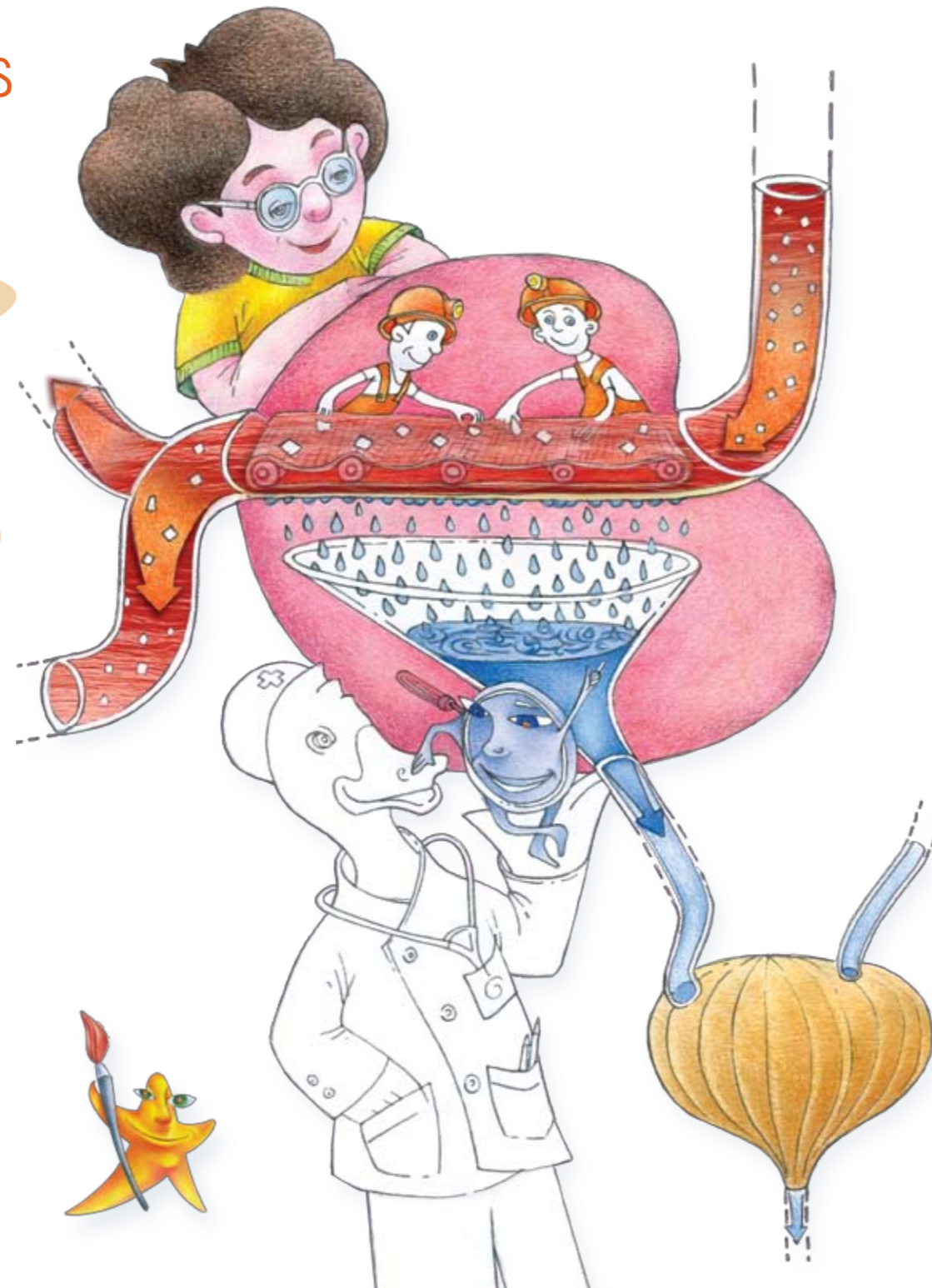
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4. The kidneys and cystinosis

You have two kidneys, one on each side of your body– they are located just under your ribs at the back and they are about the size of a fist (closed up hand).

Kidneys are very important because they do many important jobs that need to be done in a body. For example, they keep your blood clean.

Your blood is constantly moving around your body (even though you do not feel it) taking oxygen from your lungs and food and water from your tummy to where they are needed. The kidneys work to clean the blood, getting rid of waste and helping the body get rid of extra water through the wee (urine/waste water).



To do this the kidneys hold lots of the blood to clean it. In fact every drop of your blood passes through your kidneys several times a day! The kidneys work like a very good filter, filtering out all the waste. This is like a water filter that some families use to make the water from the tap clean and tasty.

So where does waste go in our body cells? You are right. It goes to the recycling centres. This includes the cells in the kidneys. If the recycling centres do not work so well (like in cystinosis), the kidneys cannot work properly. In cystinosis, this means that the kidneys cannot handle the waste and water so well – so more water and waste is lost in the wee (urine/waste water). Even waste that should have been recycled can be lost in the wee, as the recycling centres were not able to sort out the waste properly. In the waste is some of the body's salt and other chemicals that help keep us strong.



The wee (or waste water) travels from your kidneys down to your bladder. There it gets stored until it is full.

That is when you have to go to the toilet. So children with cystinosis have to go to the toilet more often to do a wee.

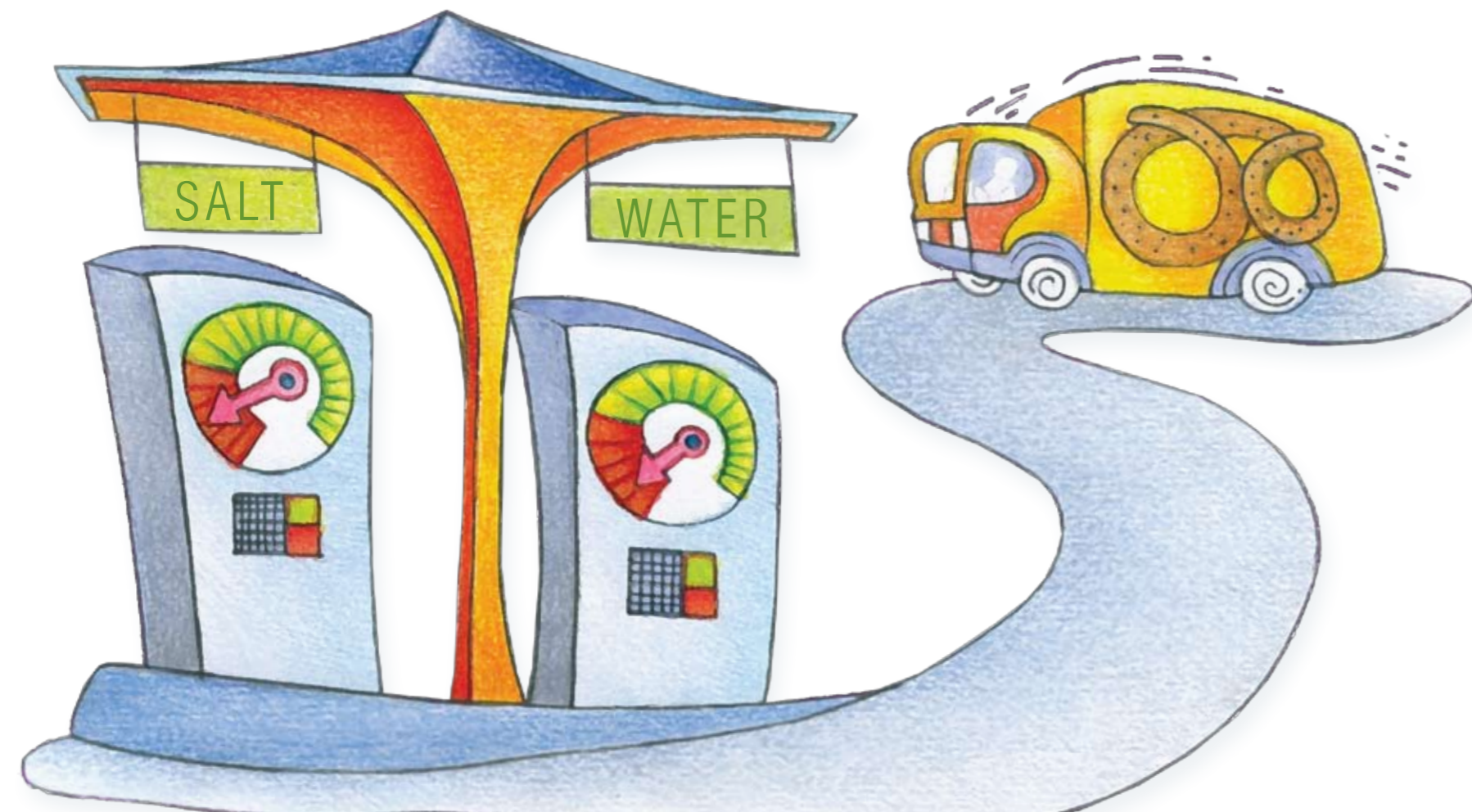
The good thing is that it is only the small recycling machines in your body that do not work – most other things work just fine! Your body knows when you lose too much water. It makes you feel thirsty and you will want to drink more.

Sometimes eating feels difficult for children with cystinosis. All the water they have to drink and the medicines they take can make it difficult to eat too.

As well as losing too much water, the kidneys in people with cystinosis lose a lot of salt that should actually stay in the blood. As well as knowing how much water you need, the body also tries to keep you with the right amount of salt you need. So people with cystinosis like salty or savoury foods to keep the salt levels right.

The good thing is that once you know that you have cystinosis, there are medicines that help the body make up for the lost salts and other medicines that will prevent the cystine from building up in your recycling centres and damaging your kidneys even more.

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One of the medicines helps to get cystine out of the recycling centres. It does this by disguising the cystine, changing its shape so it can pass through another door.

You will need to keep taking the medicine several times a day for this to work properly – your doctor will talk to you about this.

KEY MESSAGE:

Remember to take your medicine regularly so the cystine can get out of your cells and to help look after your kidneys.



Cystinosis - Where does it come from?



5. How did I get cystinosis?

Although cystinosis is very special, so only a few people have it, you are not the only one! Other boys and girls in your country and around the world will also have it. Cystinosis starts when you are growing in your mother's tummy.

The instructions that are in each cell and that make a new baby grow are called genes. Genes are what give us our hair colour, eye colour, size of nose, and other things. Genes are so small that you cannot see them with your eyes. Think of genes as like a set of instructions you get when you buy a big new toy you have to put together. You have to follow the instructions to put the toy together properly, otherwise it does not work. So the body cells have genes to help them make new proteins properly.



Do you remember when we showed you the cell earlier? All the genes are stored in the library of the cell in form of those big encyclopaedia books.

In fact we usually have two copies of each gene or instruction. You get one copy from your dad and one from your mom.

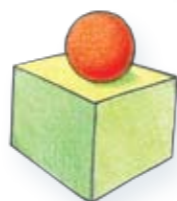
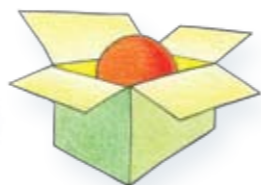
Sometimes there are spelling mistakes in the genes - just like when you make a spelling mistake at school.

Even a small spelling mistake can make a huge difference. Look at the following sentence:

Put the ball in the box

Or

Put the ball *on the box



Spelling mistakes like that mean that something in your body will not be made quite right. One of these genes tells the body how to make the cystine door in your cell's recycling centres.

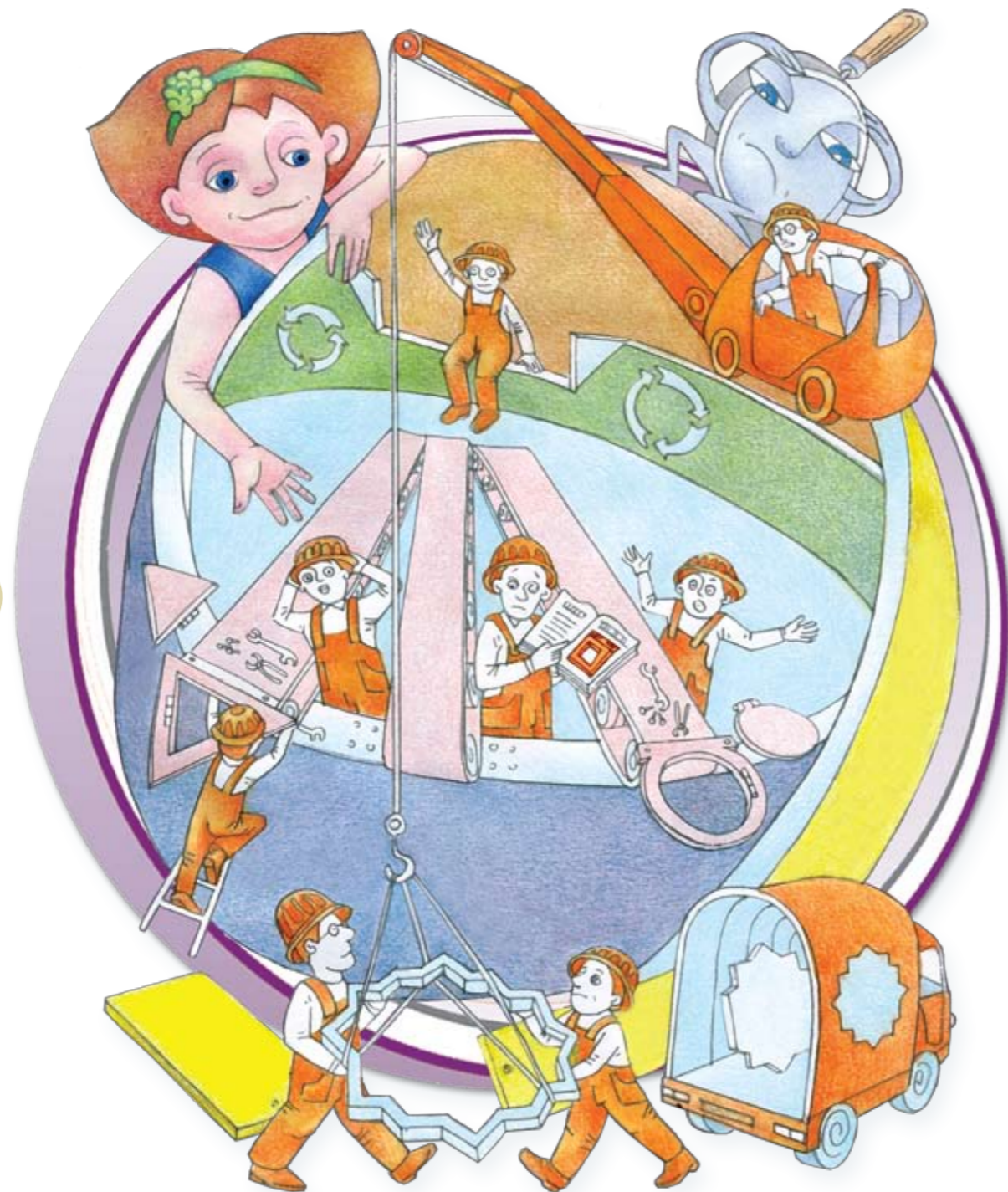
Because everyone has two copies of the gene, a mistake in one copy of the cystine door instruction does not matter because the other one will tell the body how to correctly build it. People with just one altered copy often do not even know that they have it, because they will not feel it. If both copies are not correct though, the body will not know how to build the cystine door and that person has cystinosis.



This is what happened to you. Both your mom and dad have a spelling mistake in the gene for the cystine door. When your mom and dad had you, each gave one set of instructions for your body to you. By pure chance, you got two copies with spelling mistakes in the instruction on how to make the cystine door for the recycling centre (also called a lysosome).

It is nobody's fault. Actually everyone has some altered genes, it is just bad luck if it happens. Because it is the genes that have a mistake, cystinosis is called a genetic disease; this means it is something that you are born with.

Genes and genetic diseases are a pretty complicated thing. Use this book to talk to your mom and dad or to your doctor if you need help understanding it.



6. Can you catch cystinosis from someone else?

No, you cannot catch cystinosis like you can catch a cold or a rash.

It is not due to a bug like the flu, it is something you are born with. When you feel fine, you can go to school, play with your friends and go to parties without any problem.

7. Am I different from everybody else?

Apart from having cystinosis, you have the same differences from anybody else!

Look around you! Some children are tall, some are small and your best friend might have brown hair or blue eyes. Everybody is special and there are no two people in the world that are exactly the same. Only a few people are so special that they have cystinosis.

8. When I am older, will I still have cystinosis?

Cystinosis does not go away like the measles. You will have cystinosis all your life so you will have to keep taking medicines all your life.

Cystinosis can make you feel poorly when you are young or older. So you need to keep working with your doctors and keep taking your medicines.



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But when you take them regularly, medicines will help you stay well and make sure that you can enjoy yourself with your friends and family.



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9. Will I grow up like everybody else?

Some children with cystinosis may not be as tall as other children, and they may be smaller than their friends at school who do not have cystinosis. This can happen because the kidney loses lots of salts and other things, sometimes eating is harder

and because too much cystine can make it harder for the body cells to work so well. If you have found out when you were young that you have cystinosis and you have taken your medicines, then you may be able to grow as well as many of your friends do.

Medicines



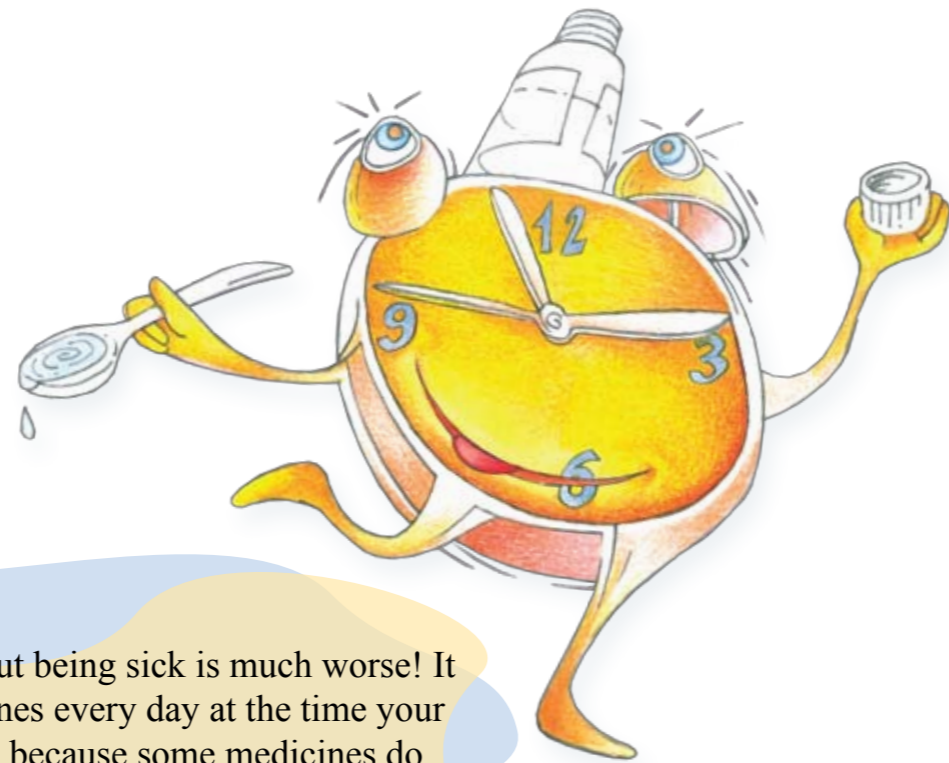
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10. Why do I have to keep taking medicines?

The medicines you take every day are helping your body. Some medicines make up for the salts and water that your kidneys are losing and others stop the cystine building up in your body so that the cells work better.

Taking medicines every day is no fun, but being sick is much worse! It is important for you to take your medicines every day at the time your doctor has told you to take them. That is because some medicines do not work for a long time and you need more. Think of it like drinking fluids. You need to drink many times throughout each day - one drink would never be enough for all day! It is the same with medicines. But not too much – take the right medicines at the right times.

Your doctor and parents can help you to remember when to take your medicines, but you are the one that has to take them! Remember how important it is for you to take them so that you stay healthy and stop the cystine building up in your body.



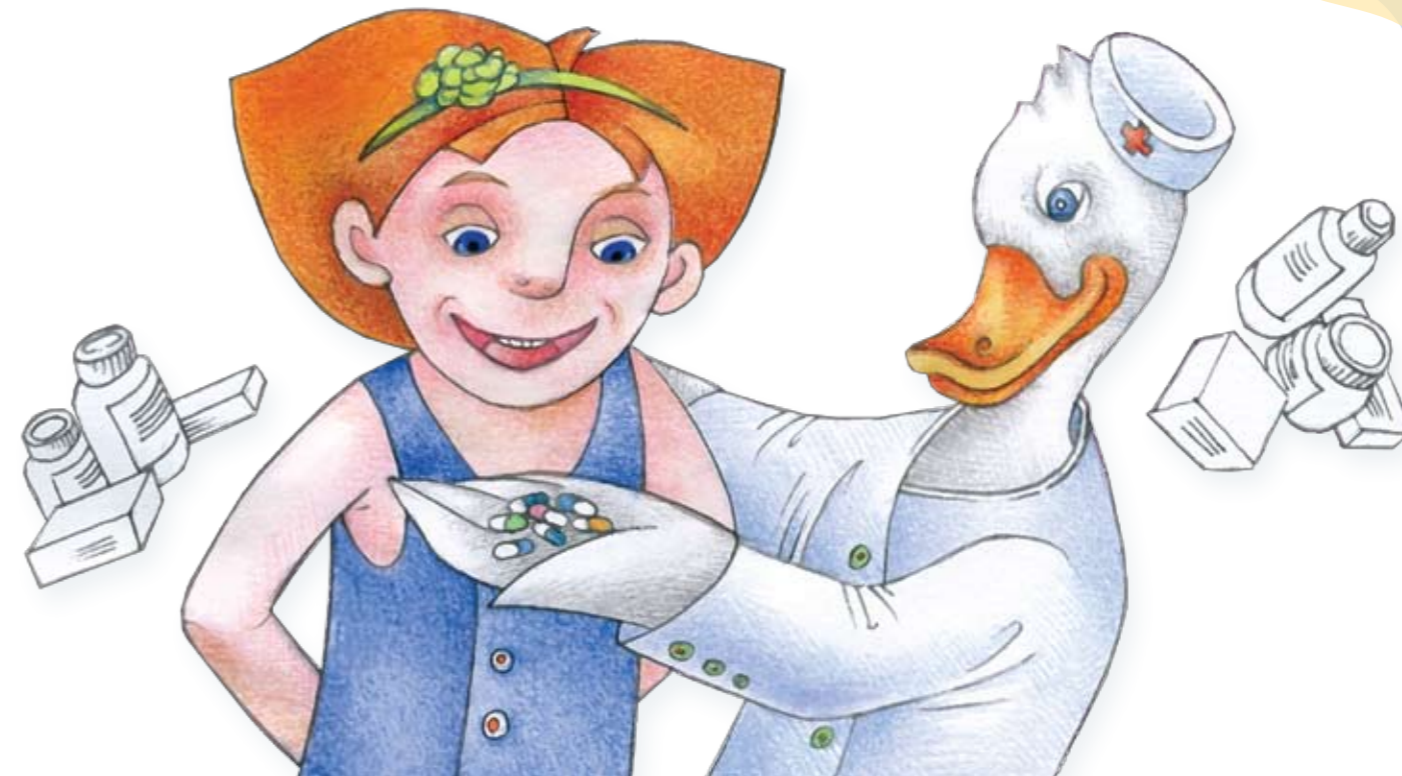
11. More about taking medicines

You will have to go on taking medicines for a long time but which medicines and how often will probably change as you get bigger and older.

Taking your medicines regularly every day will make sure that cystine does not build up too much in your cells and you can continue to do all the fun things you do today!

But, taking lots and lots of pills several times every day is also not nice – especially if some make you feel sick. And sometimes you may just forget to take them – maybe you went to a friend's house and forgot to take them with you.

We know it is not easy to take so much medication every single day, but just remember, it is what keeps you healthy!



KEY MESSAGE:
Remember to take your medications!



12. What will happen if I stop taking my medicines?



Sometimes we all forget something and that is not the end of the world. Just be careful that it happens as little as possible and that most of the time you take your medicines on schedule.

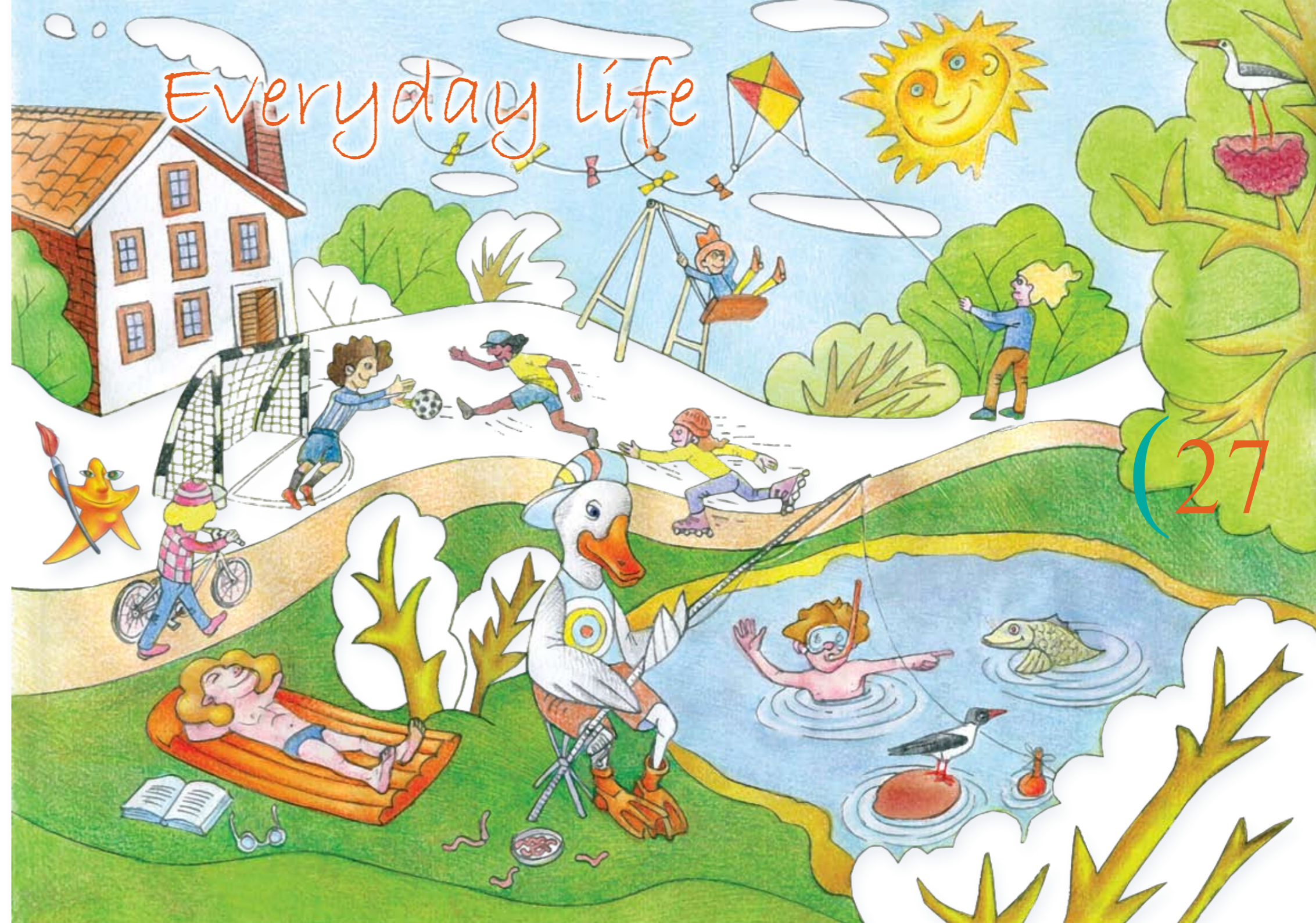
13. Where do the medicines for cystinosis come from?

Medicines for cystinosis come from the same place as every other medication. They are very carefully made in special factories.



You will not notice a change straightaway but inside your body more and more cystine will get stuck in the recycling centre with no way out. The cystine will fill up the recycling centre and it cannot do its job anymore, harming the cells. Over time, you will become sick and feel much worse.

Everyday life



14. Am I allowed to...?

Play on a computer? →

The answer is yes!

Play sports? →

The answer is yes!

Eat anything I want? →

You can eat anything your mom & dad say you can and all the yummy food they cook for the family!

Go to parties and play with friends? →

The answer is definitely yes.



Doctors



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15. Do other people need to know?

It is probably best that your school teachers know that you have cystinosis and your parents will probably talk to them about this. They will make sure you always have a bottle of water near you.

It is up to you whether or not you tell your friends. It is usually best to tell your good friends that you have cystinosis. It will help them understand why you have to take all your medicines several times each day and why you have to go to the doctor sometimes. Remember, you are the special one, you know about cystinosis and how the body cells work.

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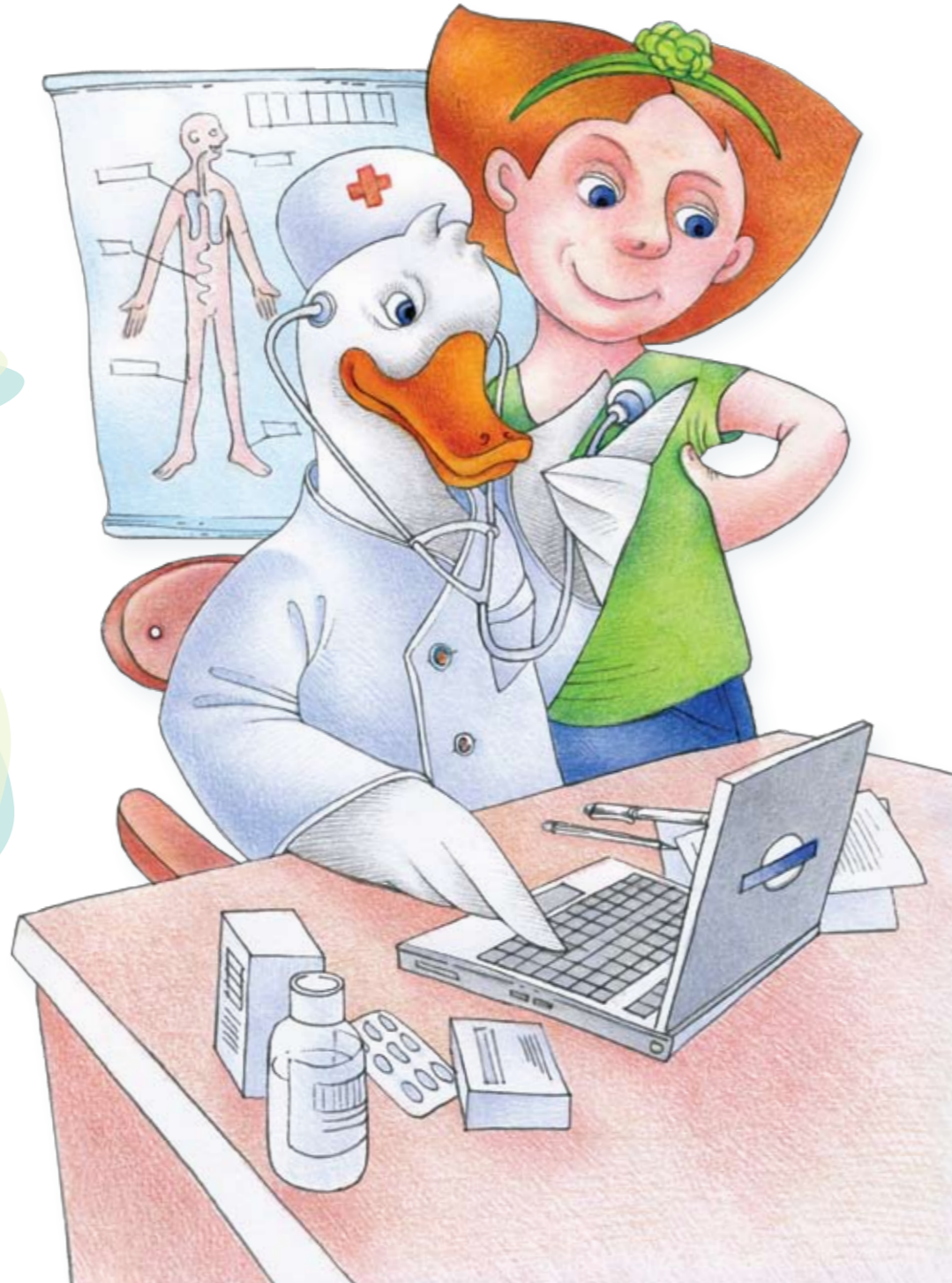
16. What does a doctor do?

Your doctor and nurse know all about the human body and about what can make you ill – but also more importantly all about everything that can make you better when you are ill such as medicines you need to take and why.

So, your doctor and nurse are there to make sure you stay well and healthy. They know all about cystinosis and about how to look after you.

But your doctor and nurse are also great people to ask any question you may have about your illness.

Some doctors help people with any sort of problem and some work mostly with children with special problems, like kidney illnesses or problems with the ways cells work. These are the doctors who will help you with cystinosis. He may be measuring how tall you are and how much you weigh. Sometimes a sample of your blood might need to be taken. Please do not be scared by this as there are things that can be done to make sure that it does not hurt. Your doctor will also need to talk to you about your medicines and whether you need to take more.



Other doctors look at your eyes. Your eye doctor will carefully examine you by looking into your eyes to see whether any cystine is present there.

17. Why are my eyes hurting when I look at the light or go outside?

Even your eyes are made of cells, and these also have recycling centres which are not working properly to remove cystine. As the medicine you swallow every day cannot get into your eyes (but pretty much everywhere else in your body), cystine will fill up the recycling centres of the cells in your eyes and will make your eyes hurt when you look at the light. You may need medicine to give as drops onto the eyes to make this go away. Your doctor will talk to you about this.

18. Why does the eye doctor look at my eyes with a special light?

Your doctor looks at your eyes with a special light in order to see whether cystine has built up in your eyes. This happens for all people with cystinosis. Medicine drops can help get rid of the cystine.





Difficult words and what they mean

Lysosomes: The cell recycling centres that have the chemicals for your body's recycling system.

Cell: Tiny little parts your body is built from.

Amino acids: Building blocks in your body's cells that come in different sizes and shapes.

Cystinosis: The name of an illness that makes the kidneys and other parts of the body ill.

Cystine: An amino acid in your body.

Kidneys: Parts of your body that filter your blood and keep it clean.

Gene: Your body's assembly instructions.

The questions and the answers



Question: How do you call the parts of your body that filter your blood and keep it clean?

Answer: Kidneys

Question: Of what tiny little parts in your body built from?

Answer: Cells

Question: What is the name for the your body's assembly instructions?

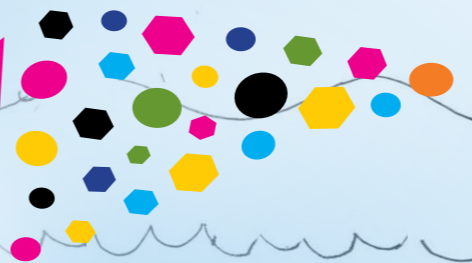
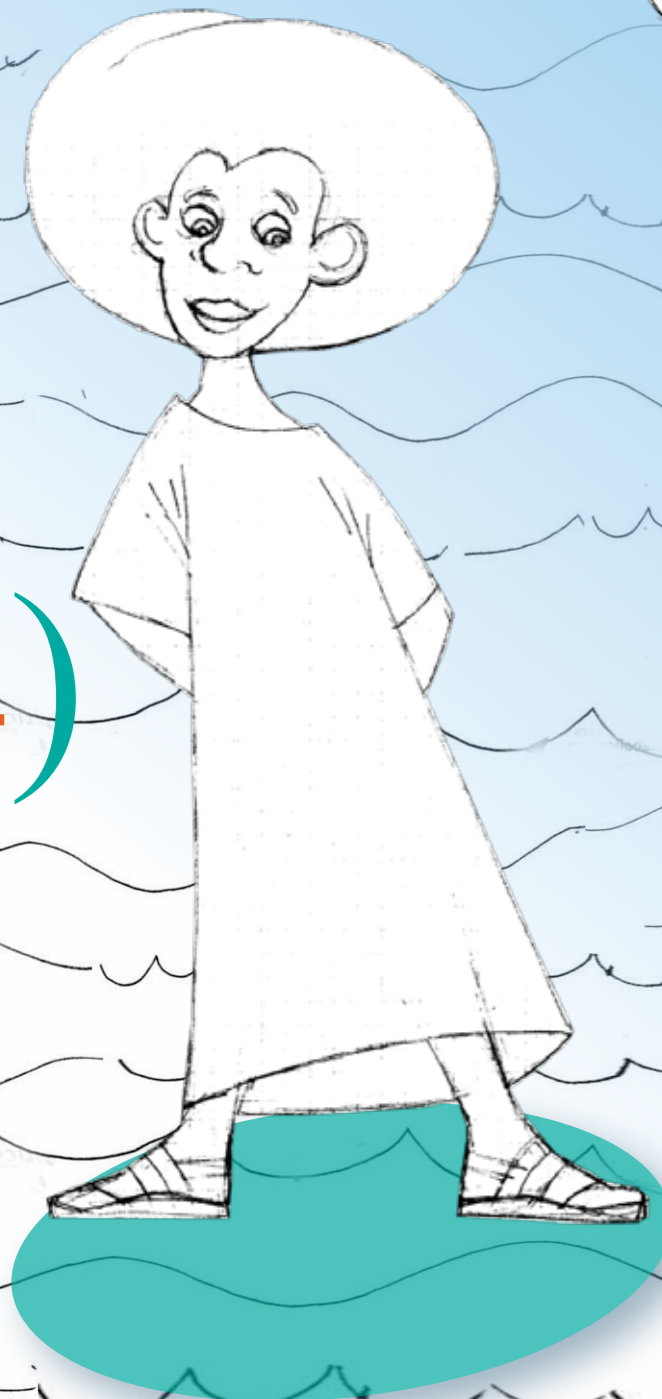
Answer: Genes

Question: What is the name of the small building blocks in your body's cells that come in different sizes and shapes?

Answer: Amino acids

Question: What part of the cell is affected in cystinosis?

Answer: The cell's recycling centre (also called lysosome)



If you have any questions regarding nephropathic cystinosis and its treatment, please contact your doctor.

This booklet was produced jointly by the **Cystinosis Foundation** and **Orphan Europe** and with the invaluable help of **Dr. William van't Hoff** “**Gt. Ormond Street Hospital for Children**”



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