An introduction to
liver glycogen storage diseases (GSD)

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Enhancing Lives Together

Foreword

This guide has been written for parents, or guardians, of children with a liver glycogen storage disease (GSD). This is because most individuals with a GSD are diagnosed in early life, when they are too young to read information about their condition for themselves. You will regularly see terms such as 'Your child', 'Your child's specialist metabolic team', etc., when referring to the person who has GSD.

If you have GSD and happen to have been diagnosed when you were a bit older, you may be reading this guide for yourself or perhaps alongside your parents. Please be assured that this guide still contains lots of useful information for you.

You can also find useful information at: **www.gsdandme.co.uk**

Always follow the guidance of your specialist metabolic team and discuss any dietary/lifestyle changes you would like to make before changes are implemented.

What are liver glycogen storage diseases (liver GSDs)?

Liver GSDs, which are sometimes called Hepatic GSDs, are a group of conditions which affect the way the body uses carbohydrate.

In this guide, we shall only be referring to the liver GSDs. We will simply call them GSD .

The liver GSDs are:

- ☐ GSD Ia
- ☐ GSD Ib
- ☐ GSD IIIa
- ☐ GSD IIIb
- ☐ GSD VI
- ☐ GSD IXa
- ☐ GSD IXb
- ☐ GSD XI
- ☐ GSD 0
- ☐ _____

- In GSD, your child has problems with the carbohydrate stores in their liver.
- Some cases are more severe than others, but all cases require lifelong management.
- GSDs are managed by following a special diet lifelong.
- With management, your child can attend school, have hobbies, enjoy sport and, as they get older, have a successful career, and enjoy a happy family life just like everybody else.

There is also a group of conditions called the muscle GSDs which are very different to liver GSDs, and they will not be discussed further here.

Diagnosis of GSD

Although GSD is sometimes picked up by routine tests carried out shortly after birth, most individuals with a liver GSD are diagnosed when they become unwell. Sometimes children with a liver GSD become unwell when they are very young babies although, depending on a variety of factors, sometimes the GSD may not become apparent for weeks, months or even years.

When GSD is suspected, the diagnosis will be confirmed through something called genetic mutation analysis testing. This will involve taking a small blood sample. In the next section, we shall look at the role genetics plays in GSD.

Why does my child have a GSD?

GSDs are inherited genetic conditions.

It's nobody's fault and there is nothing you could have done to prevent it.

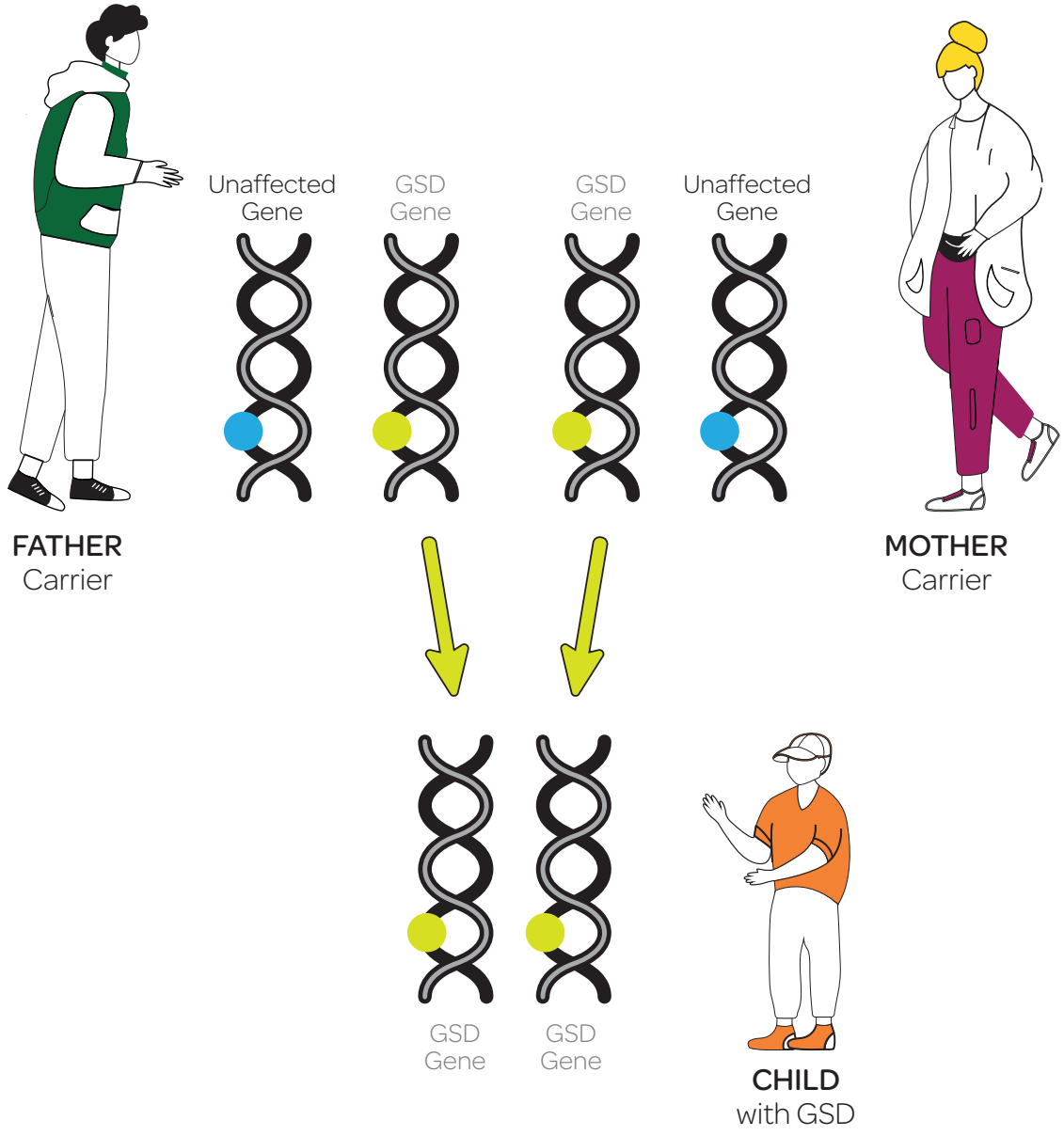
If you would like to find out how GSD type IXa is inherited, skip to pages 7 and 8.
For the other types of liver GSD, read on.

The example opposite shows how GSD is typically inherited. The scientific name for this type of inheritance is 'autosomal recessive' inheritance.

In this example, both Mum and Dad have one gene for GSD type Ia and one unaffected gene. In genetics, this is what is called being a carrier of GSD type Ia.

The child inherited two GSD type Ia genes, one from mum and one from dad.

Your child will have inherited one GSD gene from you and another gene for the same type of GSD from their other parent.


Unaffected Gene
GSD Gene
GSD Gene
Unaffected Gene
FATHER
Carrier
MOTHER
Carrier
GSD Gene
GSD Gene
CHILD
with GSD

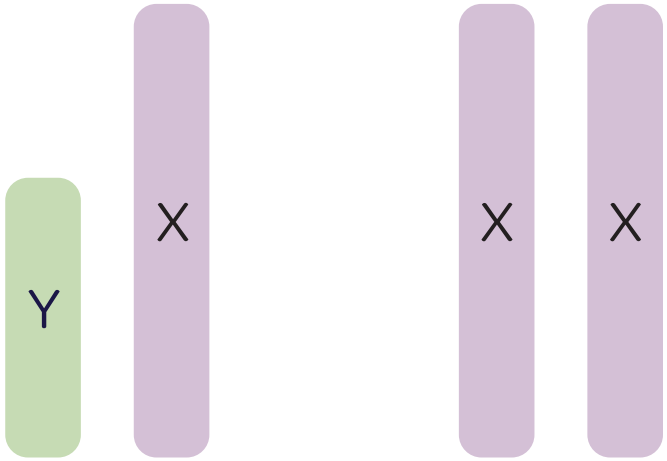
There are a small number of people who are carriers for the genes which cause GSD.

People who are carriers for a GSD gene do not have a GSD themselves and the faulty gene does not usually cause a problem to them.

GSD type IXa and X-linked inheritance

GSD type IXa is inherited in a different way. The gene for GSD type IXa is carried on what is called the X chromosome, which is one of the sex chromosomes. The sex chromosomes are the part of your genetic material that determines your biological sex.

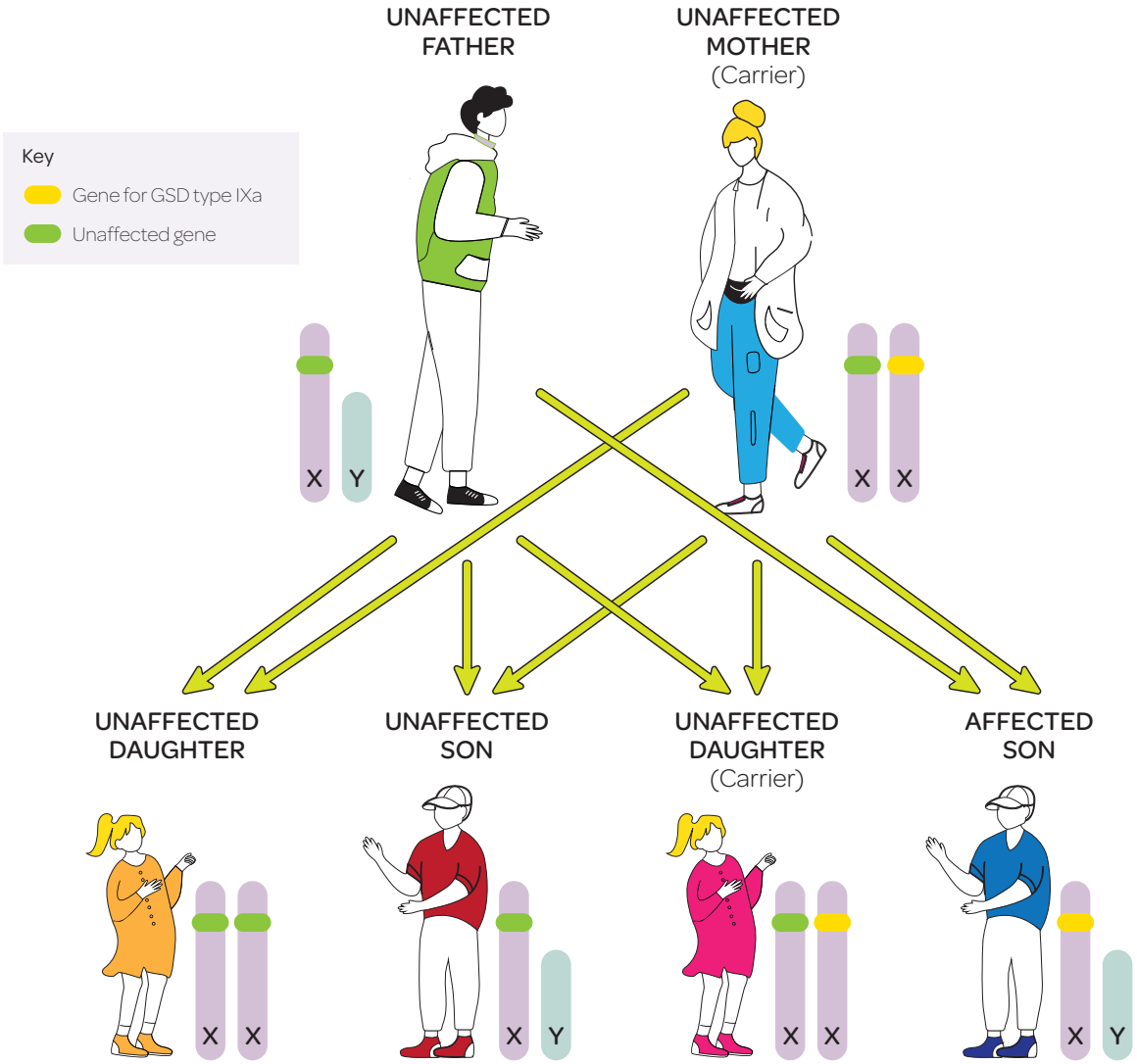
Males have an X and a Y chromosomes, where as females have two X chromosomes.



MALE FEMALE

You will notice that the X chromosome is significantly longer than the Y chromosome. This is important for GSD type IXa because the gene for GSD type IXa is carried on part of the X-chromosome that does not have a corresponding section on the Y chromosome.

This diagram shows how GSD type IXa is inherited;



GSD type IXa is carried by females but usually only males are affected. People who are carriers for a GSD gene do not have a GSD themselves and the GSD gene does not usually cause a problem to them.

Carbohydrates...The science explained

Why do we need carbohydrate?

Carbohydrate is a very important nutrient for the body.
It is a rich source of energy that our bodies needs to go about our daily activities.

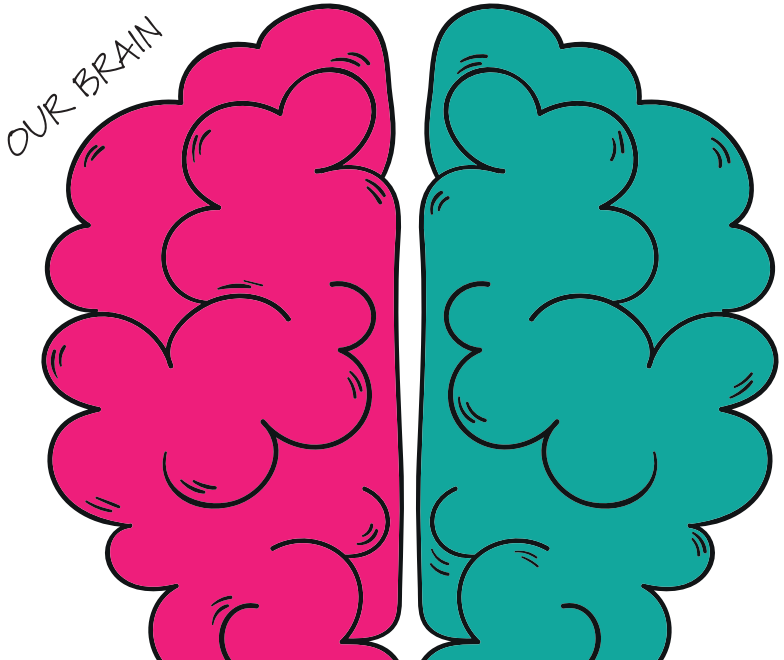

EXERCISE


DANCING


CYCLING


SWIMMING

Carbohydrate, in the form of glucose, is our brain's main energy source.



Where does carbohydrate come from?

The carbohydrate in our diets can come from many different sources.

SUGIARY FOODS

All these foods are very high in carbohydrate. These foods, however, are sometimes called 'empty calories' because they provide very little nutrition other than energy so we should not eat too many of them.

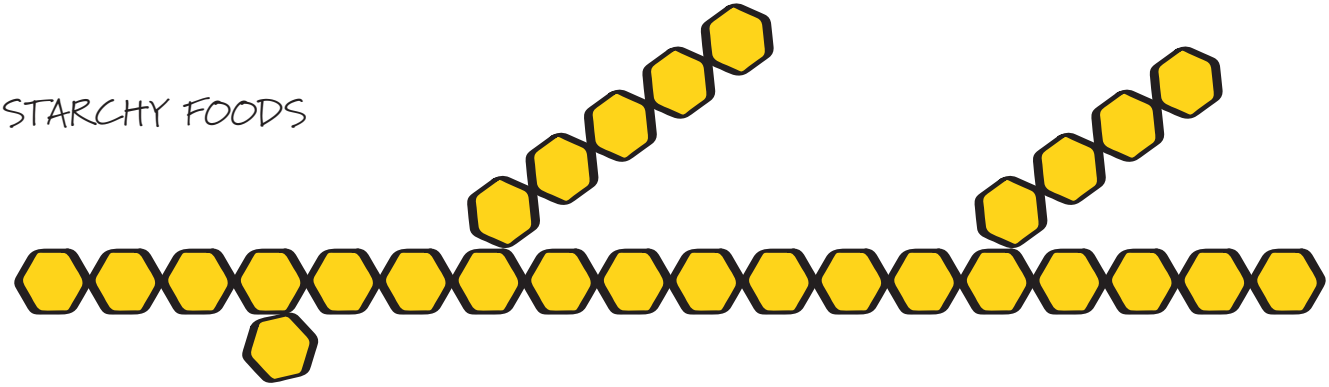

SUGIAR


SWEETS


JAM


HONEY

STARCHY FOODS



These are seen as better sources of carbohydrate, particularly the whole-grain versions, because they provide more nutrients and fibre than the sugary foods.

BREAD

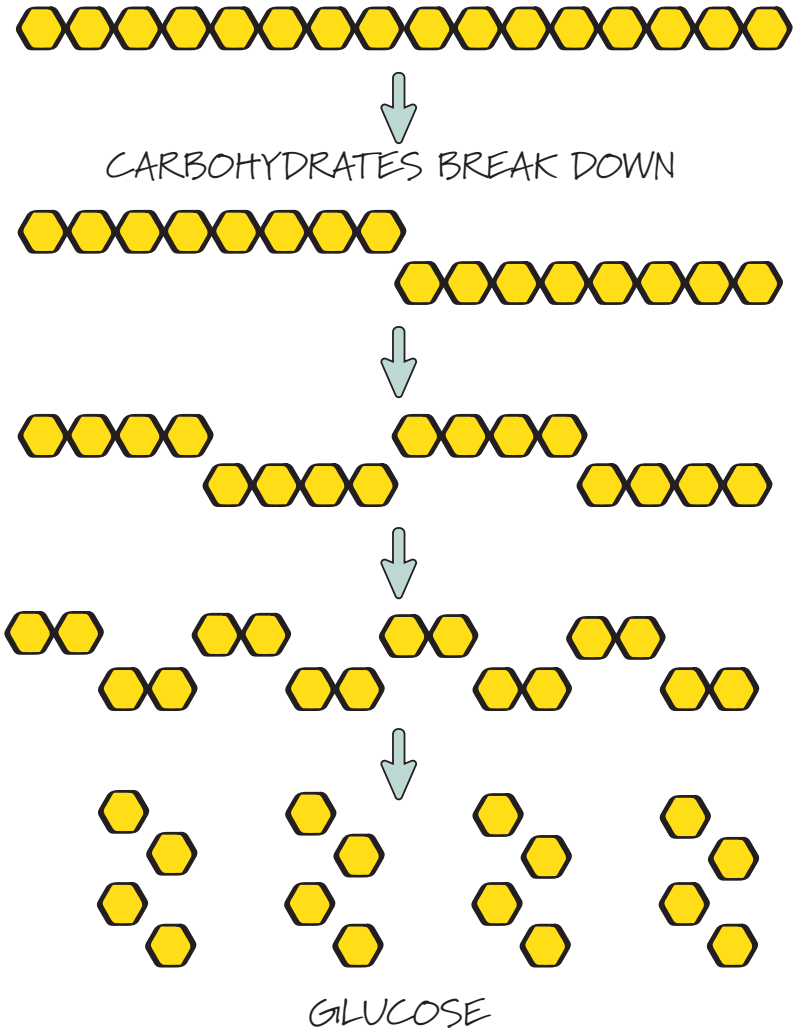
RICE

PASTA

POTATOES

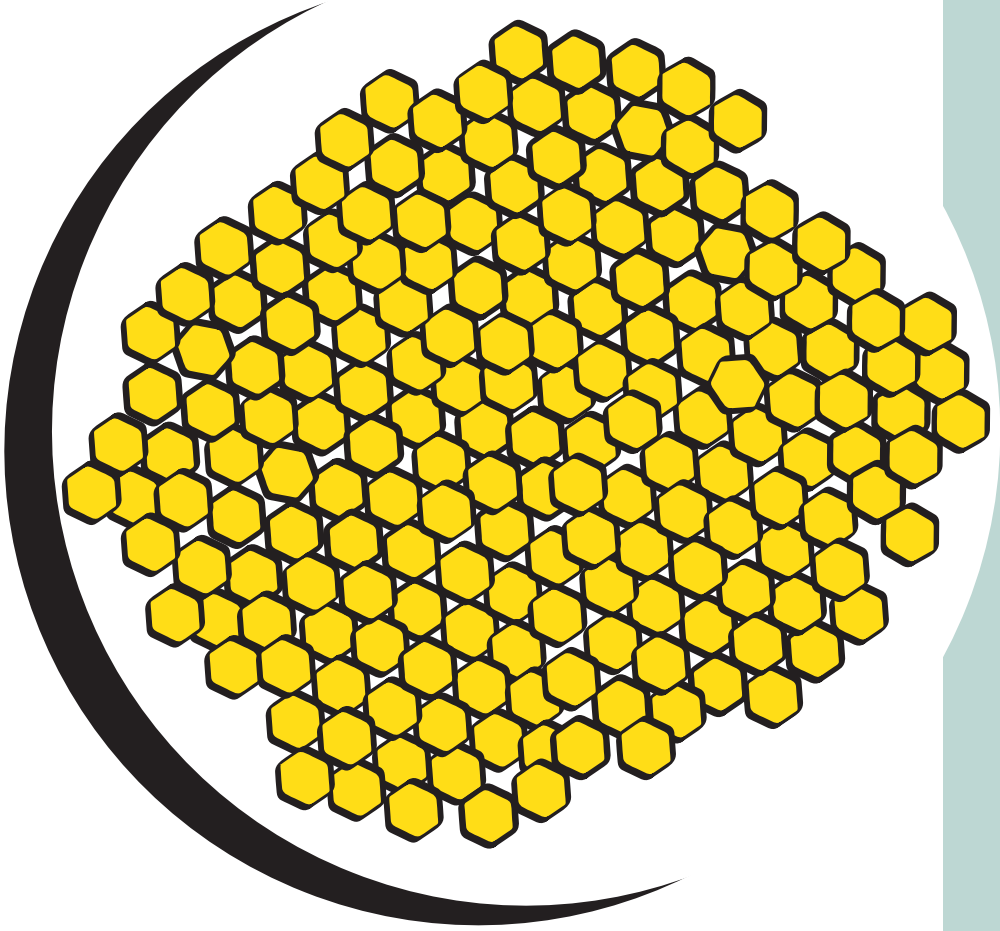
How do we get the energy from carbohydrate in our diets?

When we eat carbohydrates, our bodies break it down and we end up with the sugar, glucose. (With some foods, we can also get other sugars such as galactose or fructose but these get converted into glucose in the liver.)



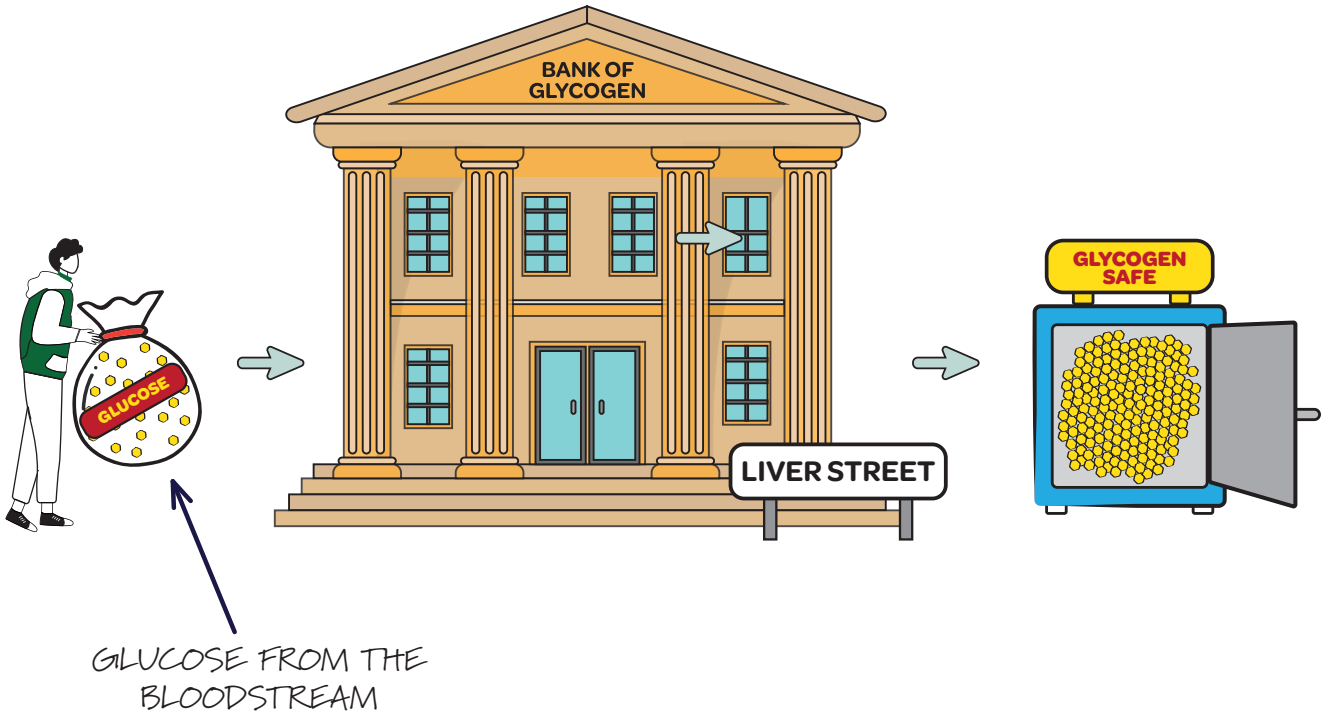
Glucose then enters the blood stream, and our cells can use it for energy. However, after a main meal, we normally have too much glucose to use all at once... so we put the glucose we don't need into our glycogen stores in our liver. Glycogen is a huge molecule made up of thousands of glucose units.

GLYCOGEN



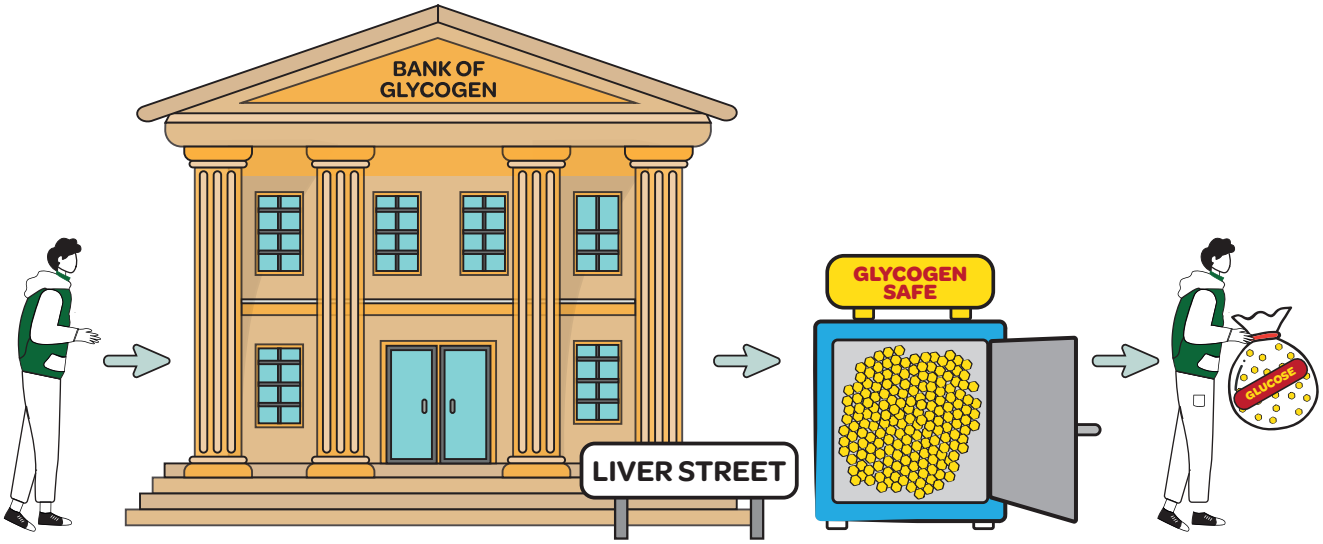
Storing glucose in the liver - Making a deposit

You can think of this like making a 'deposit' into your bank account. The glucose you are putting into your glycogen stores is like money you are saving for later use!

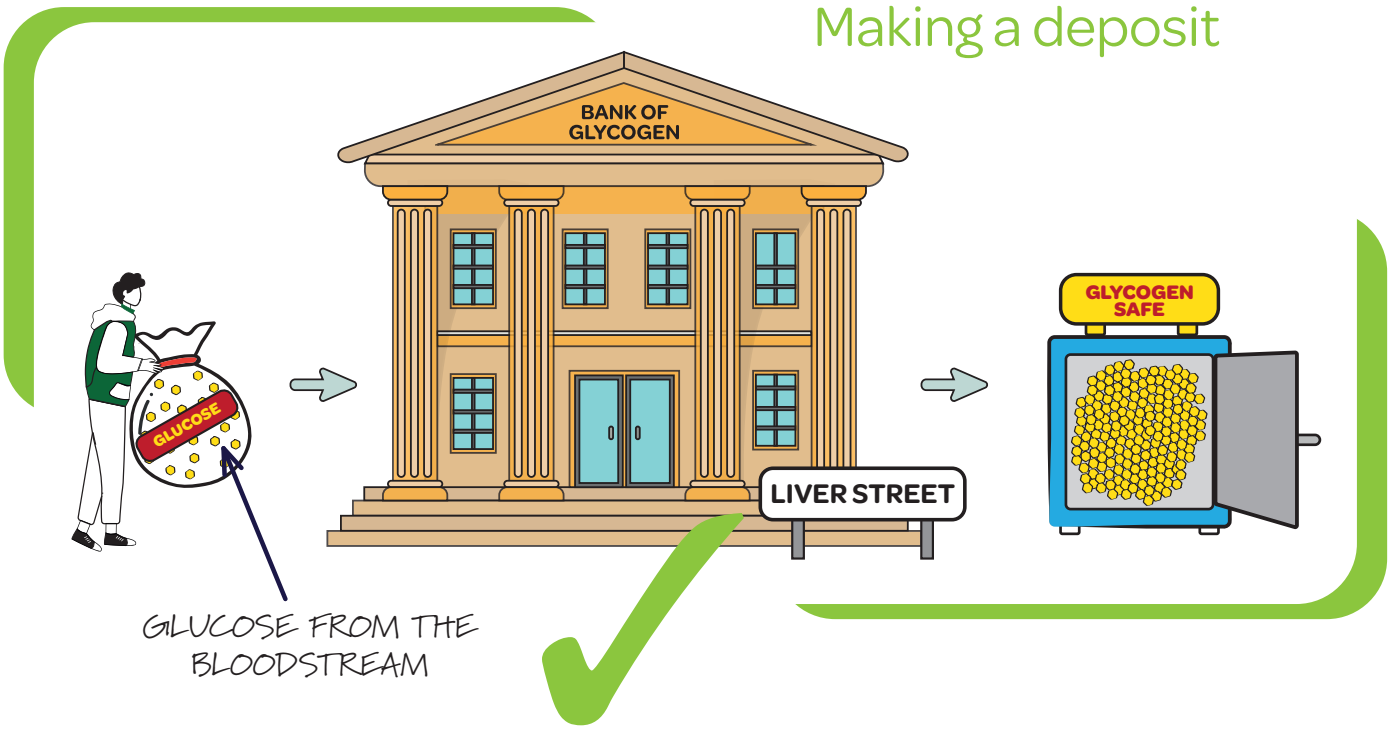


Topping up your blood glucose levels - Making a withdrawal

Within a few hours, if you don't eat again, the amount of glucose in your blood starts to decrease. Before it falls too low, your body can reclaim glucose from the glycogen stores in your liver. This is like making a 'withdrawal' of money from the bank.



So what happens in GSD?



In most liver GSDs*, what happens is similar.

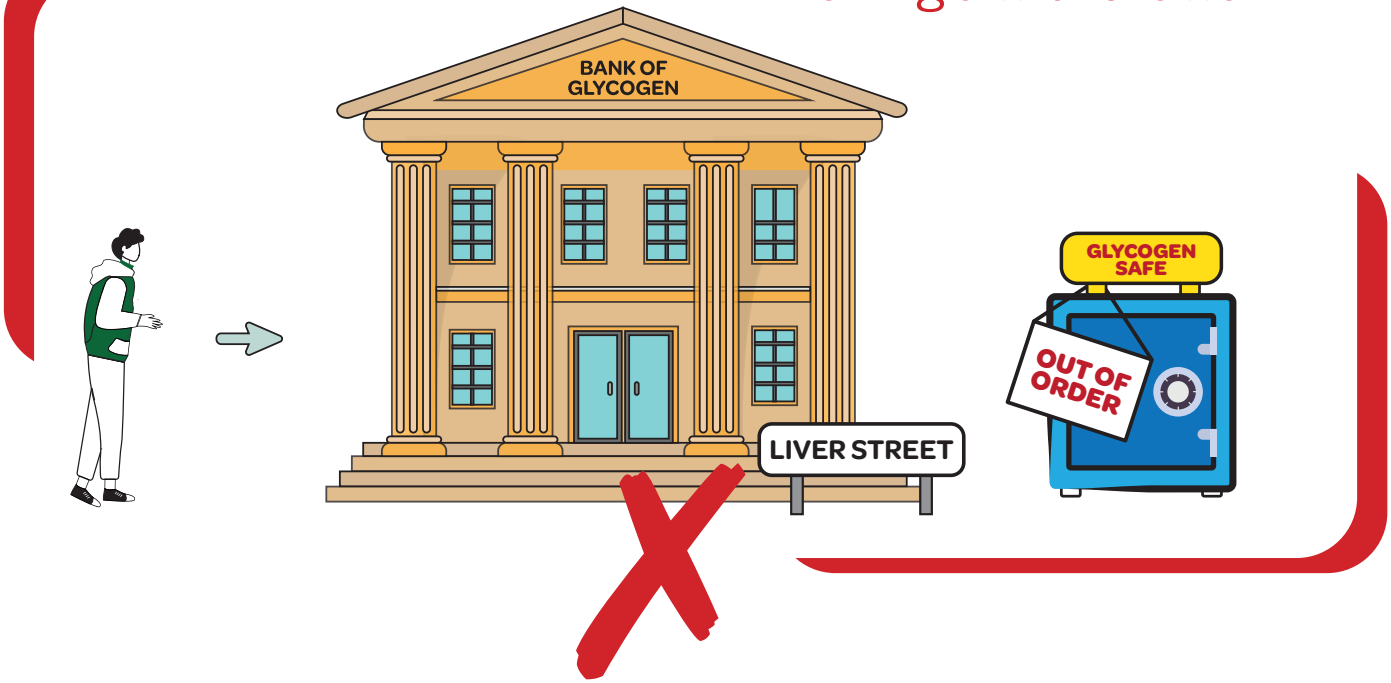
You can 'deposit' glucose into your glycogen stores but you are unable to 'withdraw' glucose from your glycogen.

This can be dangerous. It means that, just a few hours after eating, the level of glucose in your blood starts to go low. This is called hypoglycaemia or a 'hypo'.

Although some parts of your body can use fat for energy, your brain needs glucose to function properly.

Avoiding hypoglycaemia is the number one aim when managing GSD.

Making a withdrawal



With all these ‘deposits’ and no ‘withdrawals’, for some the liver can become enlarged with glycogen.

* GSD type 0 is slightly different to other types of GSD. Your child’s body cannot form glycogen and therefore they cannot even make a ‘deposit’. With no glycogen stores, the main problem is the risk of hypoglycaemia, as with the other GSDs, and management is similar.

Management of GSD

The main aim in managing your child's GSD is to keep their blood glucose level from dropping too low. Your child's specialist metabolic team will advise you on your child's safe fasting time.

The specialist metabolic team will teach you and your child how to monitor their blood glucose levels.

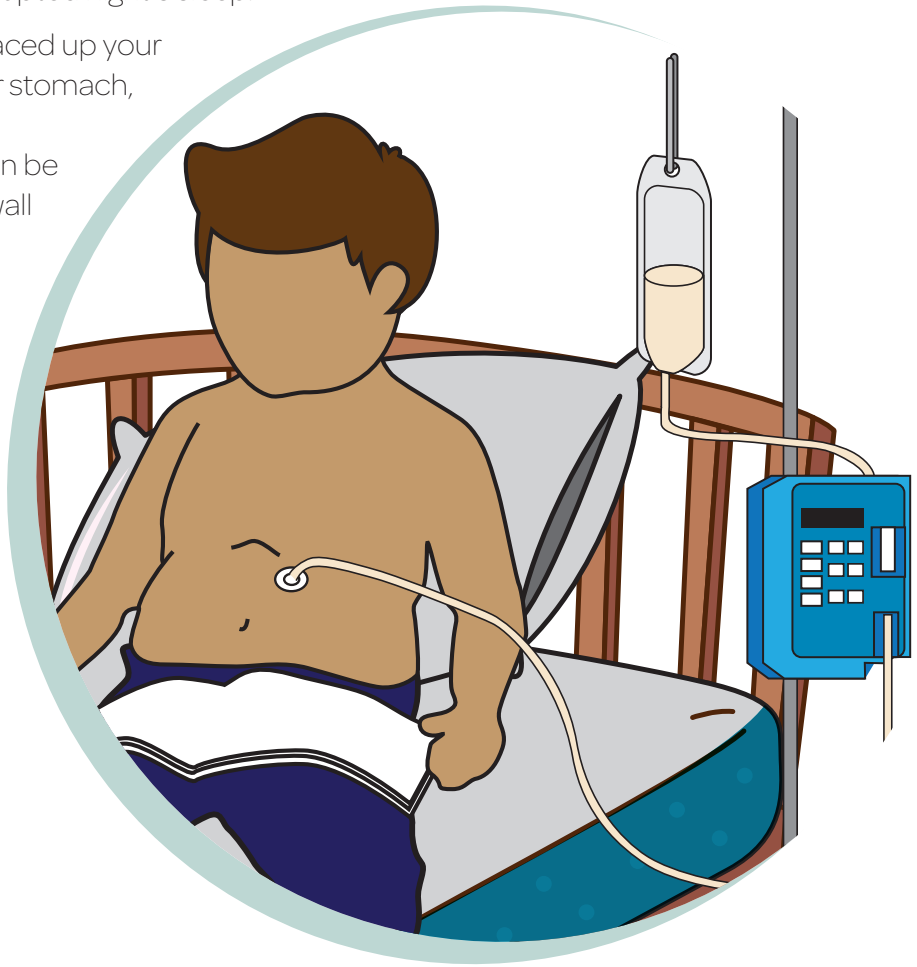
Eating regular meals and snacks will help achieve this but some people need to eat every two hours, even during the night, to prevent a 'hypo' so regular food alone is normally not enough.

Additional measures include the use of tube-feeding and cornstarch.

Tube-feeding in GSD

Tube-feeding is often used in GSD. This is where nutrition is delivered using a feeding tube. Tube feeding is mainly used during the night when your child is asleep. It provides a continuous source of carbohydrate throughout the night to maintain their blood glucose levels and prevent a hypo. This means your child (and you!) can have an uninterrupted night's sleep.

If used, a feeding tube will be placed up your child's nose and down into their stomach, this is called a nasogastric tube. Sometimes the feeding tube can be placed through their stomach wall and into their stomach, this is called a gastrostomy. Once in place, children tend to get used to the tube very quickly.



What is cornstarch and how is it used in GSD?

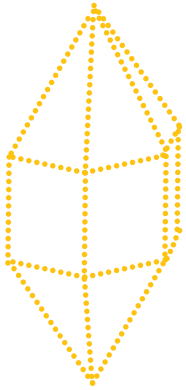
CORNSTARCH

Cornstarch, or corn flour as it is sometimes called, is an everyday cooking ingredient that can make a big difference to people with GSD.

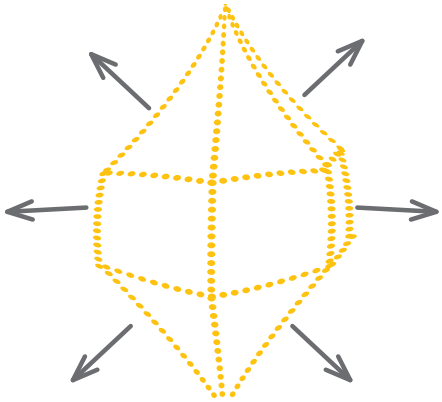
Starches are big, long chains of glucose.

When they are COOKED, like boiling a potato, starches swell up with water and become easy for the body to break down. This enables the glucose in the starch to quickly get into the bloodstream to be used for energy.

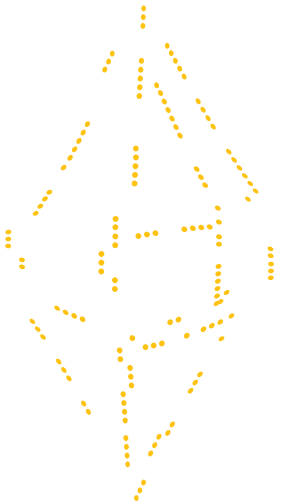
We don't normally eat UNCOOKED starches because they are difficult to break down. This is the very reason why uncooked cornstarch can be so useful in GSD – it is a slow release carbohydrate.



UNCOOKED STARCH – DIFFICULT TO DIGEST



COOKING



COOKED STARCH – EASY TO DIGEST

As cornstarch is a slow release carbohydrate, it releases its glucose into the bloodstream MORE GRADUALLY. This will help your child keep their blood glucose levels from dropping too low for longer than regular foods would.

When used, cornstarch may need to be consumed during both the day and the night.

Some doctors and dietitians do not use cornstarch before the age of two years because young children are not as good at digesting it as older children and adults are. Some hospitals will start to offer it in small amounts from a much earlier age – this will allow your child to become used to it so that, when they have to take it more regularly, they are familiar with it.

What starches are available?

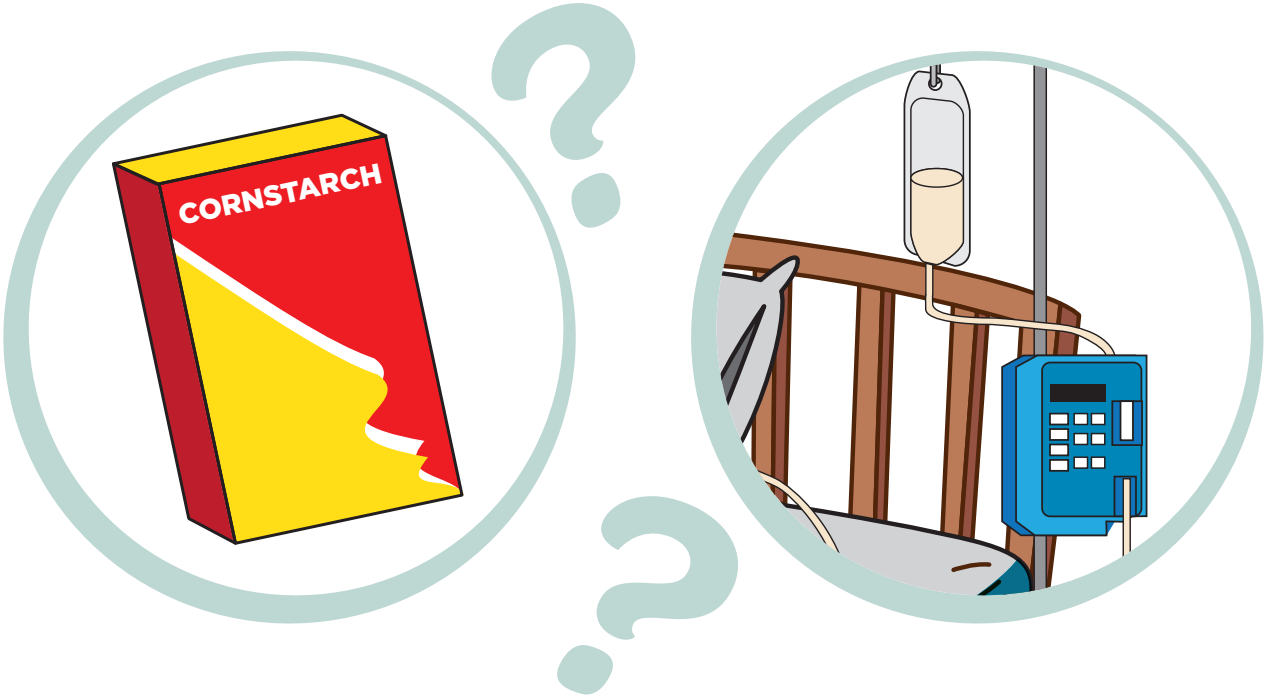
Many people obtain their cornstarch from their local supermarket. There are branded versions and supermarket-own-brand versions. In recent years, a modified cornstarch, which is only available on prescription, has become available. Your specialist metabolic team will advise on which type of cornstarch is most suitable for your child.

Cornstarch or tube feeding – which is better?

The answer to that question depends on you, your child and your family situation and lifestyle. Both can be very effective at keeping your child's blood glucose levels in the right range. The choice therefore comes down to what is best for your child. This is something that will be discussed with you by the specialist metabolic team.

Sometimes a combination of approaches is used with meals, snacks and cornstarch given during the day followed up a tube-feed at night. Sometimes a feeding tube is used to deliver the cornstarch.

As previously mentioned, cornstarch is often not used before the age of 2 years and is not suitable for young infants.



Other aspects of management

- Carbohydrate counting – We said earlier that the main aim in GSD is to avoid 'hypos' which are caused by an inability to make 'withdrawals' from the glycogen stores (bank). 'Hypos' are avoided by regularly feeding the body, through meals and snacks and/or cornstarch and/or tube feeding. However, we don't want to OVER-FEED your child either. OVER-FEEDING can cause other problems such as;

  - A build-up of glycogen stores – Too much food will cause glucose to be 'deposited' in your child's glycogen stores. We don't want this in GSD because once the glucose is 'deposited', we know it is hard for your child to 'withdraw' it and this can lead to an enlarged liver.
  - A more rapid fall in blood glucose levels than we expect! If we eat a very carbohydrate rich meal, the body responds by producing high levels of a hormone called INSULIN. High levels of insulin can cause a lot of glucose to be quickly 'deposited' into the glycogen stores. This can make the blood glucose levels drop faster than we expect which is a problem in GSD.
  - Difficulties with weight management.

Carbohydrate counting can be done in a number of different ways. The main point of it is for your child to eat an appropriate amount of carbohydrate at any one meal or snack – not too much and not too little. The specialist metabolic dietitian will advise you on carbohydrate counting.

- Nutritional supplements – With any special diet, like the diets used for GSD, your child may need to take vitamin and mineral supplements. The specialist metabolic dietitian will advise on this.

A, D, Cu, Zn

- High protein diets – In some types of GSD, high protein diets are used to maintain blood glucose levels. This might seem odd until you discover that the body can use protein that isn't being used for growth and repair to produce glucose. This is not appropriate for every type of GSD. Your child's specialist metabolic team can offer further guidance.

- Exercise – Everybody can benefit from regular exercise and there is no reason that your child cannot benefit from regular physical activity. Keeping active is thought to have additional benefits in GSD. Diet advice such as having an additional snack for times of activity may be need - please discuss with your child's specialist metabolic dietitian.

Most types of exercise will be fine for your child although contact sports (for example karate, rugby) are best avoided, particularly if your child's liver is enlarged. Their specialist metabolic team can also advise on this.

Emergency regimens

When you become unwell, your body responds in several ways. One of the things that happens is that your body starts to use up energy more quickly. This can be challenging when you have GSD - it increases the risk of a hypo. Using an emergency regimen can help minimise the impact of illness on your child.

An emergency regimen in GSD is, in its most simple form, something called a glucose polymer, mixed with water, given regularly throughout the day and night. Glucose polymers are very long-chains of glucose which are very rapidly digested. Emergency regimens keep your child's glucose levels 'topped up' throughout the day and night. The specialist metabolic team will advise you on the most suitable emergency regimen for your child.

Emergency regimens should be used at the first sign of illness. Delaying the use of an emergency regimen can be harmful to your child.

Monitoring

GSDs are life-long conditions, and your child will need to attend a specialist metabolic centre for on-going monitoring and check ups.

In early life, these visits may be quite regular but, as childhood progresses, appointments typically become less frequent.

At your child's monitoring visits, the specialist metabolic team will spend time with you to ensure your child's condition is being well managed and tailored to best suit the needs, preferences and lifestyle of your child and family.

To do this, they will look at many different factors such as:

- Your child's overall health, growth and well-being.
- The presence of any symptoms – many can be eased or eliminated through changes to dietary management.
- Blood test results – The doctors will check the results from blood tests as they can often indicate how well your child's condition is being managed.

It is very important to attend these monitoring visits. They will also give you and, as they get older, your child opportunities to discuss any concerns or questions you might have.

Closing comments

Before today, you may never have heard of GSDs. It may now seem that there is a lot of new information to take on board and that your child will have a very different diet, and perhaps a different life, to what you might have imagined.

The good news is that you are not alone. Your child's specialist metabolic team will be there to support you and your child every step of the way. They will use their experience to answer any questions you might have and help overcome any challenges that might arise. In many cases, they have encountered the same challenges you are facing with other families, and their recommendations have resulted in successful outcomes. As was said at the start, with good management, people with GSDs can live a full and active life.

Notes

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