Public Health England

NHS

Newborn blood spot screening programme

Your baby's screening result

Glutaric aciduria type 1 (GA1) is confirmed

Who is this leaflet for?

Your baby's specialist metabolic team has confirmed a diagnosis of glutaric aciduria type 1 (GA1).

This leaflet will help you understand the condition and its treatment.

What is GA1?

Glutaric aciduria type 1 (GA1), pronounced glue-ta-ric acid-ur-ee-a, is a rare but treatable inherited disorder that prevents the normal breakdown of protein.

Babies with GA1 inherit two faulty copies of the gene for GA1, one from each parent.

When we eat, our body breaks down protein in food into smaller parts called amino acids. Special chemicals found naturally in our body, called enzymes, then make changes to the amino acids so our body can use them.

Babies with GA1 have problems breaking down three amino acids called lysine, hydroxylysine and tryptophan because an enzyme is missing. This causes harmful substances to build up in their blood and urine.

Babies with GA1 benefit significantly from early treatment and can live healthy and active lives.

Without early diagnosis and treatment they can develop serious illness and damage to the brain.

Symptoms of GA1

Babies with GA1 are at risk of developing the following symptoms:

- poor feeding
- sleepiness
- floppiness
- abnormal movements
- vomiting
- breathing difficulties and fast breathing
- coldness

These symptoms can be controlled with treatment. Left untreated, GA1 can cause long-term damage to the brain, affecting muscle movement and the ability to sit, walk, talk or swallow.

When babies with GA1 become ill, they might show symptoms of metabolic crisis. A metabolic crisis is a period of time when the effects of the condition make your child seriously ill.

Some children with GA1 have been known to bleed around their brain (subdural haemorrhage). In rare cases they can develop bleeds in the back of the eyes (retinal haemorrhages).

Treatment

Treatment for GA1 involves a special low protein diet and medication.

Diet

The aim of the diet is to reduce the build-up of toxins. These toxins – glutaric acid and 3-hydroxyglutaric acid, formed from lysine, hydroxylysine and tryptophan – can cause metabolic crisis and brain injury. However, babies need a small amount of these amino acids to grow and develop. Your specialist metabolic dietitian will teach you how to measure and control the amount of protein you give to your baby, including breast milk feeds.

High protein foods and milk, including breast milk and normal infant formula, have to be limited. A special infant formula, which does not contain lysine and is low in tryptophan, is given to meet all nutritional needs.

This special infant formula is very important. It allows normal growth and development and helps reduce the build-up of the harmful toxins in the brain.

Medication

L-Carnitine helps clear some of the body's toxins. It is taken by mouth and obtained on prescription through your doctor. It should be taken regularly even when your baby is ill.

If your baby is ill

During illness babies with GA1 must be managed correctly to prevent serious physical disablities, brain damage or even death.

If your baby becomes ill or is not feeding well they should be given a special high sugar drink (also known as glucose polymer). This is called the emergency regimen. It is given without delay and frequently, day and night. Sugar free and low calorie drinks are not suitable.

The metabolic dietitian will provide detailed instructions on how to give the emergency regimen.

The emergency regimen involves stopping milk and food that contain protein. It replaces these with a glucose polymer.

It is also important to continue giving the lysine free amino acid formula. Other medications should continue while the emergency regimen is being given.

The emergency regimen is given without delay, frequently, day and night.

Sugar free and low calorie drinks are not suitable.

Your baby may need a nasogastric feeding tube for a short time during illnesses. This is a tube that goes down the nose and into the stomach. It ensures your baby receives enough regimen and medication when unable to take enough by mouth due to the underlying illness.

The specialist metabolic team will explain this to you and teach you how to use the feeding tube safely. Using the feeding tube at home might mean your baby does not have to go to hospital.

If you are worried that your baby is not improving or not taking the entire emergency regimen then you should take them to the accident and emergency department of your local hospital and contact your metabolic team.

Your questions answered

What if my baby vomits the emergency regimen drinks?

If your baby cannot keep down their emergency feeds, continues to vomit or has repeated episodes of diarrhoea despite using the emergency feeds, you should take them to hospital immediately.

You should also take your baby to hospital immediately if they seem unusually sleepy, irritable or have rapid breathing.

What if my baby needs to go into hospital?

If you have to take your baby into hospital it is important to take your copy of the British Inherited Metabolic Disease Group (BIMDG) accident and emergency guide and any medications/supplements with you.

These emergency guidelines are also available on the BIMDG website at www.bimdg.org.uk.

It is also helpful to contact your specialist metabolic team to say you are on your way, but do not delay going to hospital.

What if I lose my documents?

Contact your specialist metabolic team as they can send new copies.

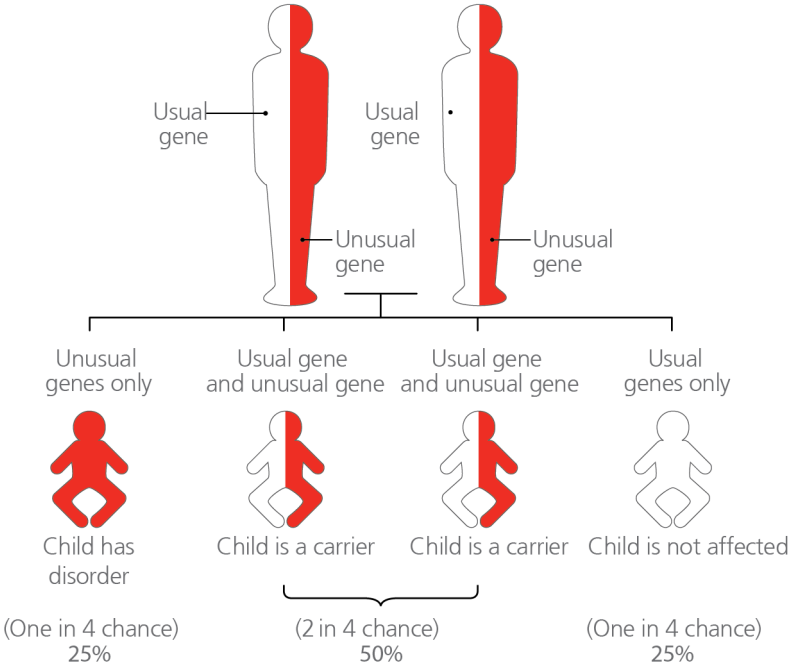
Should my other children be tested?

Your other children might be at risk of GA1 even if they have never shown any symptoms to date. It is therefore very important to get them tested if they have not been previously screened for GA1.

Your questions answered

What about future children?

A new baby from the same parents has a 1 in 4 chance of having GA1. It is important that they are tested 24 to 48 hours following birth. You should tell your midwife and GP there is a family history of GA1. You should ask for a referral to a paediatrician or genetic counsellor and make a birth plan taking their advice into account. Make sure the birth plan is written in your notes.



A small number of people carry the gene for GA1. These are known as carriers. If both parents are carriers, the baby has a 1 in 4 chance of having the condition.

Your questions answered

Who can I ask for advice and support?

The paediatric or metabolic clinician responsible for your baby's care will be happy to discuss any queries you might have.

More information and support

- CLIMB (The National Information Centre for Metabolic Diseases) provides information and support for people with GA1 and their families: www.climb.org.uk
- NHS Newborn Blood Spot Screening Programme: www.nhs.uk/bloodspot

Contact details for your specialist metabolic team:

Specialist centre	
Consultant	
Metabolic dietitian	
Clinical specialist nurse	
Ward (if applicable)	

Notes

You can write down any notes here from discussions with your health professionals

Find out how Public Health England and the NHS use and protect your screening information at www.gov.uk/phe/screening-data.

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More information: **www.nhs.uk/bloodspot**

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