



Public Health
England

NHS



Newborn blood spot screening programme

Your baby's screening result

Glutaric aciduria type 1 (GA1) is suspected



Your baby's screening result

The result of your baby's 'heel prick' screening blood test suggests they might have glutaric aciduria type 1 (GA1). A specialist team will do further tests to confirm this result.

This leaflet gives some information about GA1 and explains what happens next.



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 do not have one of the enzymes that help break down some of the amino acids. 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.

Treatment

GA1 can be treated with a special low protein diet and dietary supplements. This prevents the build-up of harmful substances.

Medication may also be given.

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

It is important that babies with GA1 feed regularly and do not go for long periods without eating. They also need to see their specialist metabolic team regularly.

If your baby is unwell in any way it is important to follow medical advice. When they are unwell they may need to go into hospital for treatment. Take any information you have about GA1 with you.

What happens next?

You have been given an appointment with a specialist metabolic team who will:

- discuss the screening test result with you
- arrange blood and urine tests for your baby
- explain how these tests can confirm if your baby has GA1

If GA1 is confirmed, the specialist metabolic team will:

- explain how to give your baby the special low protein diet. If your baby is breast fed, you can continue breast feeding
- give you any special dietary supplements and medications your baby will need for their treatment of GA1
- explain how to use an emergency feed during illness and what to do if your baby is not feeding well
- give you written information about GA1 for you to share with your family, GP and local hospital
- answer any questions you might have
- arrange a follow-up appointment to discuss the test results

If you are concerned about poor feeding or that your baby is unwell, contact your specialist metabolic team.



Your questions answered

How will I know if my baby is ill and what should I do?

Babies with GA1 do not often become poorly within the first weeks of life. However, if they have an infection, such as a high temperature or stomach upset, GA1 might cause other symptoms.

An ill baby might not feed well, be sleepy, vomit, develop breathing difficulties and become cold.

You should not ignore these symptoms. If left untreated, babies with GA1 can have fits and slip into a coma which can be life-threatening.

If you are worried that your baby is ill, contact a member of your specialist metabolic team. If you cannot contact your specialist metabolic team you should take your baby to your local accident and emergency department as soon as possible.

Take any information that you have been given about GA1 to the hospital with you.

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)	

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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information

www.nhs.uk/bloodspot



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