Sgrinio Smotyn Gwaed Newydd-anedig Cymru
Newborn Bloodspot Screening Wales

GIG CYMRU NHS WALES
Iechyd Cyhoeddus Cymru
Public Health Wales

Newborn bloodspot 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). Your baby now needs further tests to check whether he or she has GA1 or not.

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. Your baby may also be given medication.

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 will be given an appointment to see a specialist metabolic team who will:

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

If GA1 is confirmed, your specialist metabolic team will:

- explain how to give your baby the special low-protein diet (if your baby is breastfed, you can continue breastfeeding)
- give you any special dietary supplements and medications your baby will need
- 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, and
- 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, with symptoms 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

- Metabolic Support UK (The National Information Centre for Metabolic Diseases) provides information and support for people with GA1 and their families:
  www.metabolicsupportuk.org

- Newborn Bloodspot Screening Wales:
  phw.nhs.wales/newborn-bloodspot-screening

Contact details for your specialist

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

Notes

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

Using your information

For us to contact you as part of the programme, we will need to handle some of your and your baby's personal information. If you need more information about this, you can:

- visit the website: phw.nhs.wales/use-of-site/privacy-notice
- email PHW.InformationGovernance@wales.nhs.uk, or
- phone 02920 104307.

We also keep personal details to make sure that the standard of our service is as high as possible. This includes checking your baby's records if your baby is found to have a condition after having a screening test which showed a 'not suspected' result.

We only ever publish information as statistics and we never publish personal details. We pass on your personal information to health professionals or organisations that need it, including your GP, health visitor and consultant paediatrician. These professionals must protect the personal information in the same way as we do.

All our paper and computer records are stored and processed securely, and the public cannot access them.

Information courtesy of Public Health England.

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