

Isovaleric acidaemia (IVA): overview

Public Health England (PHE) created this information on behalf of the NHS. In this information, the word 'we' refers to the NHS service that provides screening.

This information is for parents if their baby is suspected of having IVA or has been diagnosed with IVA following their newborn blood spot screening test ('heel prick test'). It will help you and your healthcare professionals to talk through the next stages of your baby's care.

About IVA

Isovaleric acidaemia (pronounced iso-val-air-ik acid-ee-mia), or IVA, is a rare but treatable inherited metabolic disorder that prevents the normal breakdown of protein. Babies with IVA inherit 2 faulty copies of the gene for IVA, one from each parent.

Untreated IVA can cause long-term health problems, including brain damage, but with newborn screening and early treatment this can be prevented.

Screening and diagnosis of IVA

Newborn blood spot ('heel prick test')

When your baby was about 5 days old, your midwife took some blood from your baby's heel for their newborn blood spot screening test (the 'heel prick test'). The newborn blood spot screening test measures the amount of a substance called C5-acylcarnitine in the blood. A high level of C5-acylcarnitine suggests your baby may have IVA. This is called a screen positive result.

Diagnostic tests

If your baby has a screen positive result, you will be seen by a metabolic doctor, dietitian and nurse specialist (the 'metabolic team'). The team will provide advice and support. Blood and urine tests will be carried out to confirm if your baby has IVA.

You will need to wait a few days for the test results to be reported. During this time, you can continue to breastfeed or give normal infant formula. You should feed your baby every 3 to 4 hours, day and night, or more often if demanded. If your baby becomes unwell, you should take them to hospital for further assessment.

Some babies may already be unwell and on treatment before the screening test results are reported.

Treatment

If your baby does have IVA, the metabolic team will explain the condition in more detail and answer any questions you might have. They will start your baby on medical treatment and a special diet, and they will arrange regular follow-up appointments. Following the diet and treatment instructions is very important for your baby's health.

What to do if your baby gets ill

If your baby becomes ill, they might have an episode known as a metabolic crisis (also known as metabolic decompensation). A metabolic crisis can lead to serious illness and long-term brain damage, and can be life threatening.

To help prevent this from happening, you will be taught to give an emergency regimen, which involves specialist feeds and frequent feeding. Babies that become very unwell might need to be admitted to hospital.

The metabolic team will teach you how to look after your child during illness.

Confidentiality

The NHS Screening Programmes use personal information from your NHS records to invite you for screening at the right time. Public Health England also uses your information to ensure you receive high quality care and to improve the screening programmes. Find out more about how your information is used and protected, and your options at www.gov.uk/phe/screening-data.

More information and support

The metabolic team will be happy to discuss any queries you might have.

For more information, see www.gov.uk/government/publications/iva-confirmed-diagnosis-description-in-brief.

Further information can be found at www.metabolicsupportuk.org/. The Metabolic Support UK team can be contacted at:

- 0845 241 2173 or 0800 652 3181
- contact@metabolicsupportuk.org

The British Inherited Metabolic Diseases Group (BIMDG) website has the emergency regimen guidelines at bimdg.org.uk/site/guidelines.asp and a TEMPLE booklet about GA1 at bimdg.org.uk/site/temple.asp.

NHS.UK has information about GA1 at www.nhs.uk/conditions/glutaric-aciduria/ and newborn blood spot screening at www.nhs.uk/conditions/baby/newborn-screening/blood-spot-test/.

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