



Public Health
England

NHS



Newborn blood spot

screening programme

Your baby's screening result

Isovaleric acidaemia (IVA) 932C>T variant is confirmed



Who is this leaflet for?

Your baby's specialist metabolic team has confirmed a diagnosis of IVA 932C>T variant, also known as 'mild IVA'. This is a mild form of IVA that has very mild effects and it is unlikely that your baby will develop symptoms while well.

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

What is mild IVA?

Isovaleric acidaemia (IVA), pronounced iso-val-air-ik-acid-e-mia, is a rare but treatable inherited disorder that prevents the normal breakdown of protein. The 932C>T variant is a milder form of this disorder.

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 IVA have problems breaking down an amino acid called leucine because an enzyme is missing. This causes harmful substances to build up in their blood and urine.

The risk of problems is much lower for babies with the 932C>T variant than for those with the more serious variant of the disorder. Babies with IVA 932C>T can benefit significantly from early treatment.

Symptoms of mild IVA

When a baby with the 932C>T variant is well there are no symptoms.

Most babies will have infections with symptoms such as temperatures, coughs, colds, diarrhoea or vomiting from time to time. Babies with the 932C>T variant can usually cope with illness without difficulty.

However, as a precaution, if your baby becomes ill they will be given a special feed called the emergency regimen.

Treatment

When a baby with the 932C>T variant is well, no special treatment is needed. Your baby should not go without food or drink for long periods. No special diet or medication is required, but the dietitian will continue to monitor your baby's feed intake.

If your baby is ill

An ill baby might not feed well, be sleepy, or vomit.

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. Other medications should continue while the emergency regimen is being given.

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

Sugar free and low calorie drinks are not suitable.

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

Should my other children be tested?

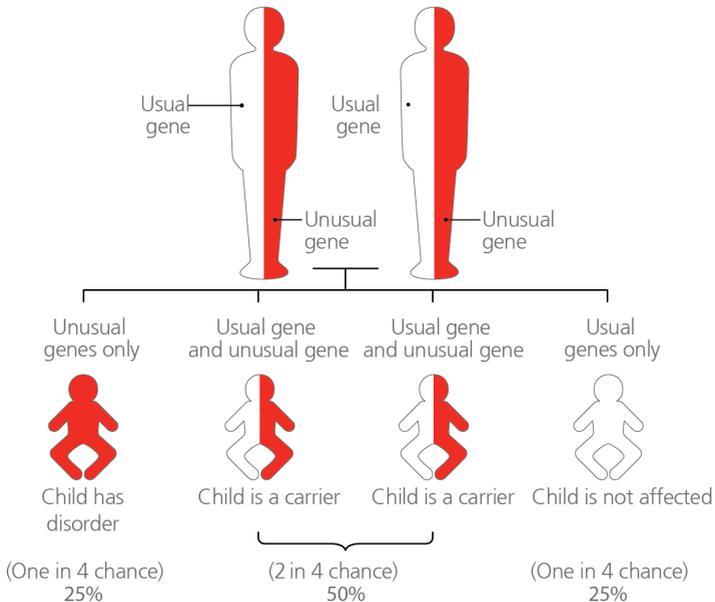
Your other children might be at risk of IVA even though they might not have shown any symptoms to date. It is therefore very important that they are tested if they have not been previously screened for IVA.



Your questions answered

What about future children?

A new baby from the same parents has a 1 in 4 chance of having IVA. 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 IVA. 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 IVA. These are known as carriers. If both parents are carriers, the baby has a 1 in 4 chance of having the condition.

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 IVA 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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More
information

www.nhs.uk/bloodspot



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