NHS
Expanded Screening Pilot

Results of the Newborn Blood Spot Screening

Parent Information Sheet Isovaleric Acidaemia (IVA) 932C>T Variant - Confirmed Diagnosis

Your baby has been seen by the Specialist Metabolic Team and a diagnosis of IVA 932C>T variant has been confirmed.
The Newborn screening test suggested IVA was present. After this finding, a special genetic test was performed to look for the presence of the 932C>T variant. This form of IVA is a mild form of the disorder and has very mild effects and it is unlikely that your baby will develop symptoms while well.

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

What is IVA?

IVA stands for Isovaleric Acidaemia pronounced iso-val-air-ik-acid-e-mia. IVA is one of a rare organic acid disorders in which a child or baby has problems breaking down protein. Protein is an essential part of our nutrition and is needed for growth and repair of tissues. In the body In order for the body to use protein from the food we eat, it is broken down into smaller parts called amino acids. Special enzymes (chemicals found naturally in your body) then make changes to the amino acids so that the body can use them.

People with IVA are missing one of the enzymes that help break down an amino acid from the foods they eat and this may cause harmful substances to build up in their blood and urine when your child is unwell.

What is the 932C>T variant?

Your baby was found to have the 932C>T variant. This change in the IVA gene results in a form of IVA with very mild effects which make it unlikely that your baby will develop symptoms.

What symptoms could we expect?

When a baby or child with the 932C>T variant is well there are no symptoms. Most babies and children will have infections with symptoms such as temperatures, coughs, colds, diarrhoea or vomiting from time to time. Babies with this variant are expected to cope with illness without difficulty. However, as a precaution if your baby becomes unwell they will be given a special feed called the emergency regimen (see below).

Does the 932C>T variant need any treatment?

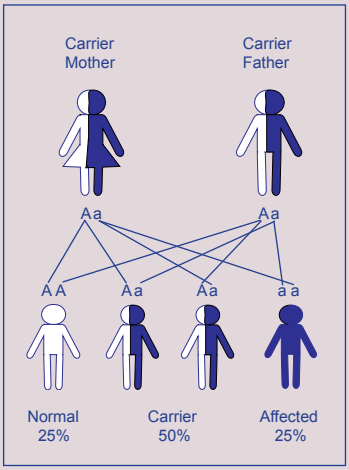
When a baby or child with the 932C>T variant is well, no special treatment is required, although it is advisable for your baby/child not to go without food or drink for long periods. No special diet or medication is required.

What happens if my baby is ill?

The Metabolic Dietitian will provide detailed instructions on how to give the emergency regimen. This involves stopping protein containing milk and food, replacing this with regular high sugar drinks (also known as glucose polymer powders i.e. SOS, Maxijul or Polycal). If your baby is not able to tolerate the emergency regimen they should be taken to hospital, so that appropriate treatment can be given. It is helpful if you contact you Specialist Metabolic Team to say you are on your way.

How will IVA 932C>T affect my child's future?

Babies with 932C>T variant IVA detected by newborn screening programmes have not experienced severe illness when unwell and have grown and developed normally.



Why does my baby have IVA?

IVA is an inherited condition. Everyone has two copies of the gene for IVA. A baby with IVA has inherited two faulty copies of the gene. The parents have one normal copy and one faulty copy and are said to be 'carriers'. When two IVA carriers have a baby, they have a 1 in 4 (25 %) chance in each pregnancy of having a child with IVA. There is nothing the parents could have done to prevent their child having IVA. The diagram shows how this happens.

Will my other children need to be tested?

Your other children may be at risk of IVA even though they may 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.

What about future children?

A new baby has a 1 in 4 (25%) risk of having IVA and it is important to test at the earliest opportunity. You should inform the Midwife and Doctor looking after you that there is a history of IVA in your family. You should ask for a referral to a paediatrician or genetic counsellor for advice then make a birth plan taking account of the information received (make sure the birth plan is written in your notes).

Who can I ask for advice and support?

The paediatric or metabolic clinician who is responsible for your child's care will be happy to discuss any queries you may have.

Contact Details for the Specialist Metabolic Team:

Specialist Centre		
Consultant		☎
Metabolic Dietitian		☎
Clinical Specialist Nurse		☎
Ward (if applicable)		☎

My notes:

Where can I find more information or support?

More information can be found at the Expanded Screening Programme website:

www.expandedscreening.org

CLIMB (The National Information Centre for Metabolic Diseases) provides information and support for people with IVA and their families.

Climb Building
176 Nantwich Road
Crewe
CW2 6BG

Telephone helpline: 0800 652 3181 (Freephone) or 0845 241 2172

Website: www.climb.org.uk

Email: info@climb.org.uk

The Expanded Newborn Screening evaluation project is funded by the National Institute for Health Research Collaborations for Leadership in Applied Health Research and Care (NIHR CLAHRC) for South Yorkshire.

We thank the UK Newborn Screening Programme Centre for their support and co-operation with the development of this booklet.

IVA 932C>T - Parent Information - Confirmed
V1.0 <27/09/12>
Not to be used after August 31st 2013
Approved by Dr Mark Sharrard,
Sheffield Children's Hospital

NHS
National Institute for Health Research