

Presumptive positive IVA**

Lab notifies IVA Clinical Liaison Service (CLS) as per local protocol on the day of the result and send original dried blood spot sample for C5 isobars

ON THE SAME DAY

1. CLS contact specialist team as per local protocol
2. Specialist team to CONTACT FAMILY to arrange urgent hospital admission
3. Specialist team to instruct family to go to specialist centre - if not possible go to appropriate hospital with 24 hr paediatric cover. Offer to arrange an ambulance
4. Specialist team to liaise with the hospital on call Paediatric Consultant (or Registrar if unable to contact) for **assessment**
 - 4a. Fax / email information to the hospital for clinicians and parents, IVA A&E letter, 'IVA is suspected' leaflet (includes NHS Newborn Blood Spot Screening Programme website address and links to parent support group), contact numbers for the IVA specialist team
 - 4b. Commence clinical management:
 - i. Well baby
 - A. Obtain diagnostic samples*** and send urgently to specialist centre laboratory (courier)
 - B. Ensure adequate feeding and obtain history of maternal antibiotics, nipple cream use and full drug history (cause of false positive)
 - C. Discharge home with BIMDG emergency guidelines and glucose polymer. Instruct to take to hospital if unwell (when baby is discharged)
 - ii. Unwell baby
 - A. Clinical assessment and admission to hospital **regardless of clinical status**
 - B. Obtain history of maternal antibiotics, nipple cream use and full drug history (cause of false positive)
 - C. Obtain blood gases, ammonia, U&E, LFT, FBC, cultures, urine ketones dipstick. Site IV cannula
 - D. Hospital to liaise with specialist team regarding clinical status
 - E. Obtain diagnostic samples*** and send urgently to specialist centre laboratory (courier)
 - F. IV 10% dextrose/0.45% saline infusion
 - G. Carnitine - specialist team to organise supply and send to local hospital if necessary
 - H. Reintroduce natural protein within 24-48 hours (refer to **dietetic management pathway******)
 - I. Transfer to specialist centre as soon as appropriate
5. Specialist team to liaise with diagnostic laboratory - inform lab to expect samples (including transport arrangement) and which hospital child has gone to in case samples need following up
6. Hospital to feedback to specialist team with a review within 2 hours of admission if not already transferred to specialist centre
7. Specialist team to inform GP (as soon as practicable), send IVA GP letter via fax / email
8. Specialist team to inform maternity services and health visiting services

FIRST REVIEW with parents within 24 hours of screening result
If not already seen in specialist centre, speak directly via telephone or other communication
 Review available test results
 Reintroduce natural protein within 24-48 hours, refer to dietetic management pathway****
 (If inpatient at DGH, do not discharge until agreed by specialist team)

WELL BABY
 Specialist team to review face to face within 5 working days of diagnostic sampling with the results

UNWELL BABY
 Specialist team to review face to face as soon as practicable with the available results. Arrange to feedback remaining results by 5 working days of diagnostic sampling

Plasma C5 ≥cutoff
 Urine IVG normal
 Urine MBG normal

Plasma C5 ≤cutoff
 Urine IVG normal
 Urine MBG normal

Plasma C5 ≥cutoff
 Urine IVG normal
 Urine MBG increased

Plasma C5 ≥cutoff
 Urine IVG increased
 Urine MBG normal

Uncertain
 Repeat investigation

Effect of antibiotics (Pivalate)
 Check pivalate results from initial screening sample

Screened false positive
IVA UNLIKELY
 No treatment needed - discharge

Short/branched chain acyl-CoA dehydrogenase deficiency (SBCAD)
 Long term FU
 No ER

IVA CONFIRMED
 Treat as IVA (refer to dietetic and clinical management guidelines)
 Arrange sibling screening if IVA confirmed*****

* See IVA initial clinical referral guidelines and standards for further details

** See IVA screening protocol for details

*** See IVA diagnostic protocol for confirmatory test details

**** See IVA dietetic management pathway on BIMDG website - www.bimdg.org.uk/site/guidelines-enbs.asp

***** See IVA sibling protocol for details

Treat as IVA
932C>T allele detected

- Feed normally
- Glucose polymer emergency regimen for intercurrent illness
- See dietary management guidelines****
- Weaning/post-weaning FU visits and others as needed

932C>T allele not detected

- Start/continue with carnitine (100mg/kg/day in 2-4 divided doses) and/or glycine (150mg/kg/day divided in 3 doses) supplementation
- If clinically unwell consider protein restriction. See dietary guidelines****
- Emergency regimen