

Initial C5 > 5.0 μmol/L (immediate referral before isobar results), or Initial C5 ≥ 2.0 and ≤ 5.0 μmol/L and Isobar analysis consistent with IVA → **Presumptive positive IVA\***  
 Initial C5 ≥ 2.0 and ≤ 5.0 μmol/L and Isobar analysis consistent with other condition → **Other condition suspected\***

Lab notifies IVA Clinical Liaison Service (CLS) as per local protocol on the day of the result

**Presumptive positive IVA: 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 hour 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
5. 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 of the IVA specialist team
6. Commence clinical management:
  - a. Well baby
    - i. Obtain diagnostic samples\*\* (bloodspot acylcarnitine, urine organic acids, DNA sample) and send urgently to specialist centre laboratory via courier
    - ii. Ensure adequate feeding and obtain history of maternal antibiotics, nipple cream use and full drug history (potential cause of false positive)
    - iii. Discharge home with BIMDG emergency guidelines and glucose polymer. Instruct to take to hospital if unwell.
  - b. Unwell baby
    - i. Clinical assessment and admission to hospital regardless of clinical status
    - ii. Obtain history of maternal antibiotics, nipple cream use and full drug history (potential cause of false positive)
    - iii. Obtain blood gas, ammonia, U&E, LFT, FBC, cultures, urine or blood ketones (point of care analysis) & Site IV cannula
    - iv. Hospital to liaise with specialist team regarding clinical status
    - v. Obtain diagnostic samples \*\* (bloodspot acylcarnitine, urine organic acids, DNA sample) and send urgently to specialist centre laboratory via courier
    - vi. Commence IV 10% dextrose/0.45% sodium chloride infusion
    - vii. Commence levocarnitine 100mg/kg/day (Specialist team to advise and organise supply and sent to local hospital if necessary)
    - viii. Reintroduce natural protein within 24-48 hours (refer to dietetic management pathway\*\*\*\*)
    - ix. Transfer to specialist centre as soon as possible
7. 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
8. Hospital to feedback to specialist team with a review within 2 hours of admission if not already transferred to specialist centre
9. Specialist team to inform GP (as soon as practicable), send IVA GP letter via email
10. Specialist team to inform maternity services and health visiting services.

**Other condition suspected: On the same day**

1. CLS contact specialist team as per local protocol
2. Follow above steps as per local specialist team guidance for assessment but do not use IVA information as IVA not suspected

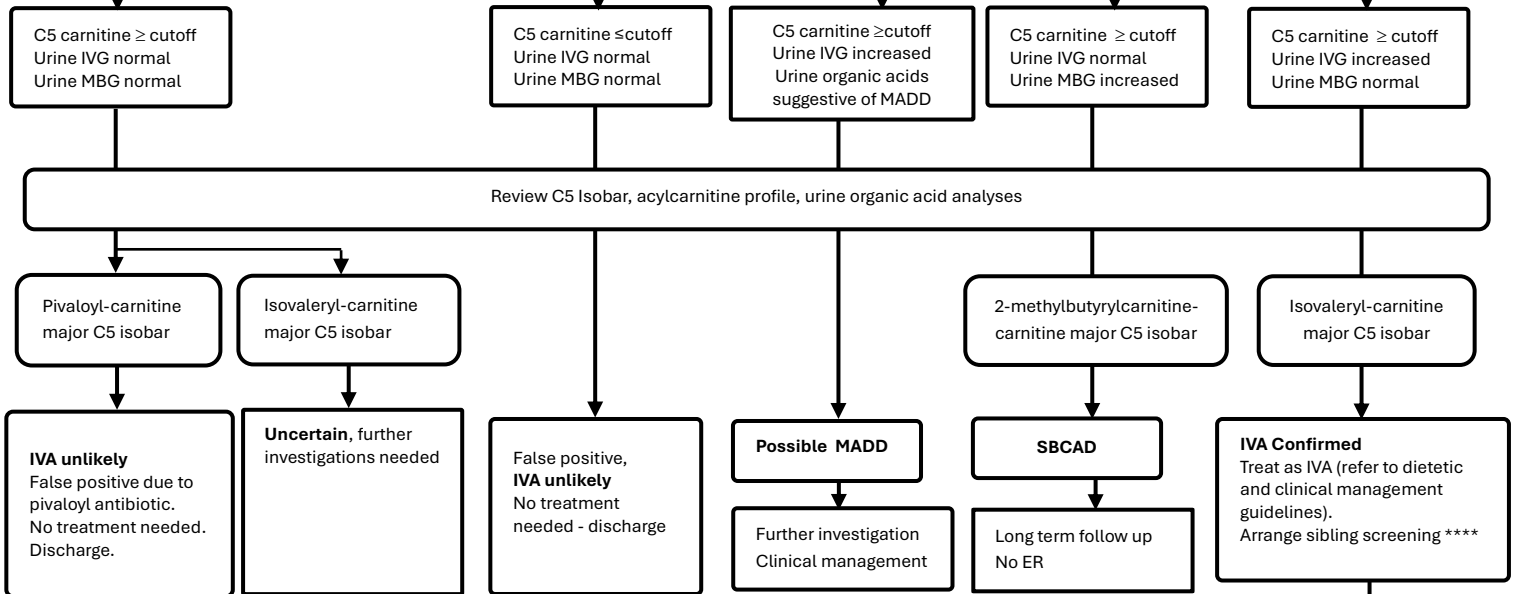
**First review with parents within 24 hours of screening result**  
 If not already seen in specialist centre, speak directly via telephone or other communication  
 Review available test results.  
 Reintroduce natural protein within 24-48 hours, refer to dietetic management pathway\*\*\*\*  
 (If inpatient in local hospital, 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



\*See IVA screening protocol in IMD Lab Handbook for details  
 \*\* See IVA diagnostic protocol in IMD Lab Handbook for details  
 \*\*\* See IVA dietetic management pathway on BIMDG website  
 \*\*\*\* See IVA sibling protocol in IMD Lab Handbook for details

**Abbreviations:**  
 IVA – isovaleric acidemia  
 IVG – isovalerylglycine  
 MBG – 2-methylbutyrylglycine  
 SBCAD – short/branched chain acyl-CoA dehydrogenase (2-methylbutyryl-CoA dehydrogenase) deficiency  
 MADD – multiple acylCoA dehydrogenase deficiency  
 ER – emergency regimen

**Treat as IVA**  
**932C>T allele detected**

- Feed normally
- Glucose polymer ER for intercurrent illness
- See dietary management guidelines\*\*\*
- Weaning/ post-weaning follow-up visits and others as needed

**932C>T allele not detected**

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