

MCADD clinical management guidelines*

Presumptive positive MCADD**

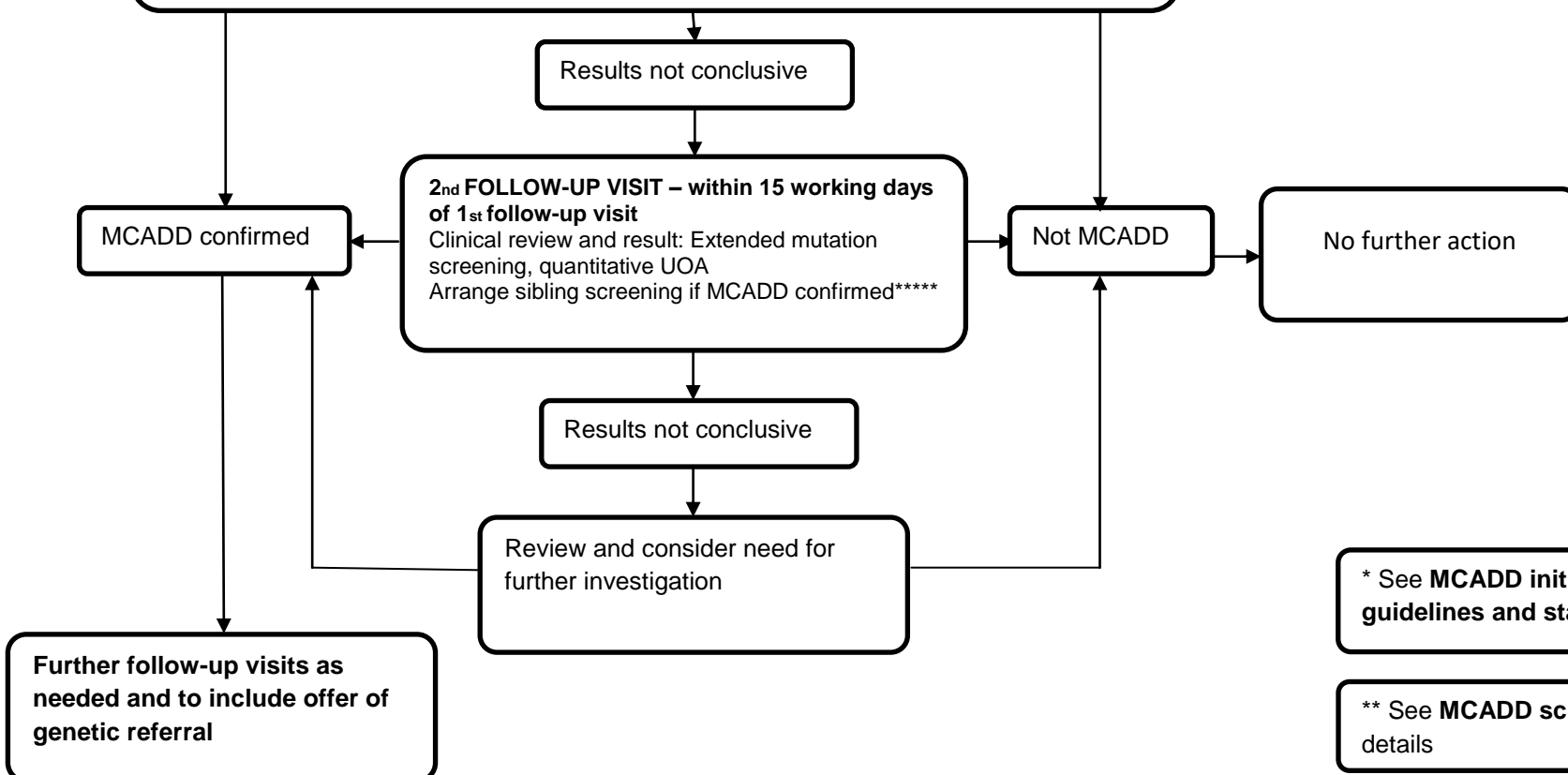
Lab notifies MCADD Clinical Liaison Service (CLS) as per local protocol on the day of the result.

ON THE SAME DAY

1. CLS contact specialist / designated team as per local protocol. The following information MUST be provided to the clinical team by the screening laboratory:
 - (a) Hospital of Birth
 - (b) Parent telephone number
 - (c) Telephone number of the midwife
2. Specialist / designated team to CONTACT FAMILY. If family CANNOT BE CONTACTED, see ♦♦♦ at the bottom of this page.
 - (a) If infant well and feeding regularly:
 - a. Advise parents about regular feeding and emergency management of MCADD if the infant becomes unwell, including assessment at the local hospital, as per (b) below. Arrange face-to-face hospital review either the same day or the next working day, as per local arrangements.
 - b. Arrange for the family /parents to receive the 'MCADD is suspected' leaflet and MCADD A&E letter, and contact details of the specialist team.
 - c. Contact the paediatric team at the local or specialist hospital (as per local protocol) to inform them of the patient, and make arrangements for assessment and management if the infant becomes unwell.
 - (b) If infant unwell/feeding poorly/doubts about clinical situation
 - a. Specialist team to instruct parents to go to local hospital or specialist hospital A&E (as per local protocol)
 - b. Specialist team to liaise with the hospital on call Paediatric Consultant (or Registrar if unable to contact) for assessment and initial management
 - c. Fax / email information to the hospital for clinicians and parents, GP letter, MCADD A&E letter, 'MCADD is suspected' leaflet (includes NHS Newborn Blood Spot Screening Programme website address and links to parent support group), contact numbers for the MCADD specialist / designated team and location
 - (c) FIRST REVIEW: FACE-TO-FACE – (same day or next working day, as per clinical status and local arrangements):
 - a. Well baby
 - i. Obtain diagnostic samples*** including DNA and send urgently to diagnostic laboratory
 - ii. Ensure adequate feeding and dietetic review**** (Emergency regimen and feeding review, advice on safe fasting times)
 - iii. Ensure family have specialist / designated team contact details
 - iv. Discharge home with A&E letter, appropriate leaflet (MCADD is suspected / MCADD is confirmed), emergency regimen (ER)****, BIMDG emergency guidelines**** and glucose polymer. Instruct to take to hospital if unwell
 - b. Unwell baby
 - i. Obtain diagnostic samples*** including DNA and send urgently to diagnostic laboratory
 - ii. Follow BIMDG emergency guidelines****
 - iii. Before discharge, ensure adequate feeding and dietetic review
 - iv. Ensure family have specialist / designated team contact details
 - v. Discharge home when well enough with A&E letter, appropriate leaflet (MCADD is suspected / MCADD is confirmed), emergency regimen (ER)****, BIMDG emergency guidelines**** and glucose polymer. Instruct to take to hospital if unwell
 - (d) Follow up to be arranged at specialist / designated centre within 5 working days and inform GP, send MCADD GP letter via fax / email

First follow up visit – within 5 working days of first face-to-face contact.

Clinical review including dietetic review and emergency regimen
 Review results: Octanoylcarnitine (C8), qual. urine organic acid (UOA), c.985A>G mutation analysis
 If MCADD is confirmed***, review dietetic advice and emergency regimen ****. Arrange sibling screening if there are unscreened siblings*****
 If diagnosis not confirmed – see diagnostic protocol***



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

** See MCADD screening protocol for details

*** See MCADD diagnostic protocol for confirmatory test details

**** See MCADD dietetic and emergency guidelines on BIMDG website: www.bimdg.org.uk/guidelines/guidelines-child.asp

***** See MCADD sibling protocol for details

♦♦♦ If family cannot be contacted directly, consider the following options:

- Leave message on phone and ask to call back. Phone again after 2 & 4 hours if no contact
- Ascertain if baby in hospital of birth & contact appropriate paediatric consultant / registrar
- Contact midwife or community midwifery team for information via labour ward where baby born
- Home visit – if specialist nurse/midwife available; if not at home, leave note asking family to make contact
- Attempt contact/visit again following day