

HCU Clinical Management Guidelines*

Presumptive positive - HCU**

Lab notifies HCU Clinical Liaison Service (CLS) on the day of the result

ON THE SAME DAY

1. CLS contact specialist team as per local protocol
2. Specialist team to CONTACT FAMILY and arrange appointment next working day. Provide family with 'HCU is suspected leaflet' either via web link or in person. *(Parents should NOT be informed of a positive result if an appointment cannot be given for the same or next day e.g. parents should not be informed on a Friday unless an appointment is for that day or the Saturday. In this case contact should be deferred until after the weekend to the next working day).*
3. Specialist team to inform GP (as soon as practicable), send HCU GP letter via fax / email
4. Specialist team to inform maternity services and health visiting services

FIRST REVIEW: FACE-TO-FACE - (same day or next working day)

Obtain diagnostic samples***
 Give supply of pyridoxine 50 mg bd & folic acid (5mg/day) to be started if diagnostic tests confirm positive for HCU

Within 5 working days of diagnostic samples:
 Review diagnostic results available
 Communicate results to family

HCU CONFIRMED

No

Diagnostic test negative (false positive)

HCU UNLIKELY
 Explain results to the family and discharge baby

Yes

Repeat plasma homocysteine and amino acids (local hospital or specialist centre to be organised)
 Start pyridoxine 50mg bd & folic acid 5mg/day

Repeat plasma amino acids and total homocysteine after taking pyridoxine and folic acid for approximately 1 week and again after approx 2 weeks (results within 2 working days).
 Discuss dietary treatment and provide family with Methionine-free formula. See HCU dietetic management pathway****

Pyridoxine responsive (homocysteine in target range*)

Yes

Communicate with family. Continue pyridoxine and folic acid. Doses may be adjusted

No/Partial response*

Communicate with family. Start methionine restricted diet - refer to HCU dietetic management pathway****
 Continue folate. Continue pyridoxine if there was partial response. Doses may be adjusted

Clinical Management via specialist team
 Arrange sibling screening*****

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

** See HCU screening protocol for details

*** See HCU diagnostic protocol for confirmatory test details

**** See HCU dietetic management pathway details on BIMDG website

***** See HCU sibling protocol for details