

PKU clinical management guidelines*

Presumptive positive PKU**

Lab notifies PKU Clinical Liaison Services (CLS) on the day of the result

1. ON THE SAME DAY

- a. 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
2. PKU CLS contact designated (or specialist) team as per local protocol to arrange 'first review' appointment (to take place by next working day).
3. PKU CLS ensure appropriate face-to-face contact is made with family that day (nurse specialist/GP/HV/midwife as per local protocol).
4. PKU CLS to fax/email following information to GP: GP letter, 'PKU is suspected' leaflet, contact details for PKU designated (or specialist) team, appointment time and location.
5. Ensure family given above information (except GP letter) when first family contact is made.

FIRST REVIEW: FACE-TO-FACE - (within 24 hours of informing parents of the screening result)

Take diagnostic samples***

Dietetic review and commence dietary management if appropriate **within 24 hours******

Clinician to ensure:

- i) Family have received – PKU designated (or specialist) team contact details and appropriate dietary guidelines
- ii) Letter have been sent to: GP, local paediatrician, local dietitian as necessary

1st REVIEW OF AND COMMUNICATION OF DIAGNOSTIC RESULTS – within 15 working days of 1st face-to-face review

Clinical review and results: phenylalanine, tyrosine, pterins and dihydropteridine reductase (DHPR)

Arrange sibling screening if PKU confirmed*****

If diagnosis not confirmed – see diagnostic protocol***

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

**See PKU screening protocol for details

***See **PKU diagnostic protocol** for confirmatory test details

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

*****See **PKU sibling protocol** for details

PKU confirmed

Yes

Follow-up visits for on-going management
Dietetic and clinical review

No

Follow diagnostic protocol***