Homocystinuria (HCU) Dietetic Management Pathway

Presumptive positive screen for HCU
Refer to Clinical Management Guidelines and Initial Clinical Referral Guidelines and Standards (www.bimdg.org)

1st Follow-up visit: after 14 days of pyridoxine and folic acid
Repeat plasma amino acids and total homocysteine
Discuss dietary treatment, provide family with methionine free formula eg: HCU Anamix Infant (as per local protocol)
Blood results 2 working days (results communicated to family)

Pyridoxine unresponsive / partial response
commence dietary treatment
- Methionine restricted diet
- Continue 50mg pyridoxine/day 5mg folic acid/day if partially responsive

Pyridoxine responsive
Continue pyridoxine and folic acid.
No diet

Breast feeding regimen
Restrict breast milk intake to approx 50% by giving methionine free formula (approx 75mls/kg/day) prior to breast feeds (mature breast milk contains ~18mg methionine/100mls)
(expect plasma tHc and methionine to decrease to target treatment range 7-12days)
Review acceptance of methionine free formula bottle feeds - within 24 hours. If not accepting bottle then temporarily stop breast feeds to establish methionine free feeds. Follow bottle feeding regimen guidelines.
Support mum to express milk.

Bottle feeding regimen
Decrease or temporarily stop normal formula according to plasma methionine levels
Give methionine free formula only depending on plasma methionine at 1st follow-up visit

Plasma Methionine µmol/L	Methionine free formula only number of days
>1000	4
600-1000	3
200-600	2

Reintroduce whey based infant formula (contains ~ 29mg methionine/100ml)
Limit methionine intake initially to 90-120mg methionine/day, (or 30mg/kg/day if weight <3kg)
Supplement with methionine free formula to give total protein intake 3g protein/kg/day

Dietetic review ~ 1 week on dietary treatment
Monitor plasma methionine and tHc ~ 7days after methionine free feeds started (results in 2 working days)
Adjust volume of methionine free formula to maintain tHc and methionine within biochemical targets.
Breast milk intake/infant formula typically provides around 90 - 120mg methionine/day.

Plasma target treatment levels:

methionine	normal range
total Hcy	60 - 100 µmol/L
free Hcy	≤ 10 µmol/L
total cysteine	normal range

Monitor blood levels, weekly initially and adjust feeding regimen to achieve target treatment levels

Inform GP, health visitor, local dietitian, as appropriate, of patient management plan

Homocystinuria - Dietetic Management

Dietary management information (as per local centres own resource)

- HCU Dietary information
- Weaning information
    - low protein 'free' foods
    - low protein manufactured foods
    - basic 20mg methionine / 1g protein exchanges
    - calculating protein exchanges
    - low protein prescribable products
    - low protein weaning recipes
    - second stage L-amino acid supplement
- Prescription letters to GP
    - methionine free amino acid supplements
    - low protein manufactured foods & advice regarding prescriptions

- Home delivery services

Follow-up in first year - 'suggested' dietetic management for HCU

Follow-up visits	Dietetic management pathway
1 week following introduction of diet	Wt, length (weekly weights) Review feeding – volume of standard infant formula (methionine intake), volume methionine free formula – check total protein intake ~ 3g protein /kg/d Review feeds with results of repeat plasma total homocysteine levels, adjust methionine intake (from infant formula or breast feeds) as necessary to achieve biochemical target levels Monitor plasma cysteine: 1-2 monthly. Supplement if < 170 µmol/L
Weekly telephone reviews	Review feeding regimen with results total plasma homocysteine and methionine levels, adjust feeds as necessary to achieve target levels Once achieved target levels consistently (over 4 – 8 weeks) could change to fortnightly monitoring & weights.
4 Months	Wt, length (consider monthly weights) Review feeding – volume standard infant formula (methionine intake), volume methionine free formula – check total protein intake ~ 3g protein /kg/d Weaning stage 1 • Discuss low protein 'free' baby foods
6 Months	Wt, length (monthly or more as necessary) Review feeding – volume standard infant formula (methionine intake), volume methionine free formula – check total protein intake ~ 3g protein /kg/d Weaning stage 2 • expand range of low protein free foods, include prescribable low protein foods • discuss introduction of methionine containing foods (teach methionine / 1g protein exchanges) to gradually replace breast / normal infant formula feeds • gradually introduce second stage methionine free L- amino acid powder (as paste or add to methionine free infant formula).
8 Months	Wt, length (monthly or more as necessary) Weaning stage 3 • expand range of low protein free foods include prescribable low protein foods • continue introduction of methionine containing foods (exchanges) • introduce more texture/finger foods into diet (as per usual weaning practices). • introduce more family low protein meal choices • increase second stage methionine free L- amino acid powder/decrease methionine free formula
12 Months	Wt, length (monthly or more as necessary) Weaning stage 4 • family foods, encourage more variety, • Give advice re: toddler feeding