**TITLE: NURSING GUIDELINES FOR THE MANAGEMENT OF CHILDREN**

**WITH CLASSICAL PHENYLKETONURIA**

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| **REVISION HISTORY** | | | |
| POLICY TITLE: NURSING GUIDELINES FOR THE MANAGEMENT OF CHILDREN WITH  CLASSICAL PHENYLKETONURIA | | | |
| DOCUMENT REFERENCE NUMBER: PP-CLIN-NUR-109 | | | |
| Revision | Active Date | Status | Comment |
| Rev. 2 | 23/01/2018 | Obsolete |  |
|  | | | |

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| --- | --- |
| **METHOD OF COMMUNICATION /DISTRIBUTION:** | **QPULSE & EMAIL** |
| RESPONSIBILITY FOR IMPLEMENTATION /TRAINING: | Eilish O’Connell, Clinical Education Facilitator, NCIMD |
| RESPONSIBILITY FOR EVALUATION AND AUDIT: | Eilish O’Connell, Clinical Education Facilitator, NCIMD |

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# **PURPOSE:**

The objectives in preparation of Nursing Guidelines for Management of Inherited Metabolic Disorders (IMD) are to increase the knowledge base of nursing staff involved in the delivery of care to patients with an IMD, provide a resource material for reference and ultimately ensure the consistent delivery of high quality care to patients attending the National Centre for Metabolic Disorders (NCIMD).

**Readers of this document are reminded that prescription of dietary regimes and all medications (including insulin, minerals, vitamins and trace elements) is the responsibility of the Metabolic Consultant. These guidelines may only be used under the supervision and guidance of a Metabolic Consultant.**

*The document authors wish to thank the various Doctors, Nurses, parents and patients who have worked in and attended the National Centre throughout the years, contributing greatly in the process to our knowledge and experience of Inherited Metabolic Disorders.*

# **DEFINITIONS:**

PKU is the most common inborn error of protein metabolism. The incidence in Ireland is 1:4,500 (Naughten, 1996). Phenylalanine is an amino acid that cannot be metabolised to tyrosine due to the absence or deficiency of the liver enzyme Phenylalanine Hydroxylase. Elevated phenylalanine levels are neurotoxic and if left untreated result in intellectual disability. However, following the introduction of Newborn Screening in 1966 in Ireland, along with early dietary intervention the natural course of this condition has altered (Clarke, 2006).

**INCIDENCE**

Ireland 1: 4 500 (Naughten et al, 1996)

Worldwide 1:12 000

USA 1:14 000

Japan 1:60 000

Finland 1:1,000,000 (Walter et al, 2012)

**INHERITANCE**

PKU is an autosomal recessive disorder (i.e. both parents are carriers of the defective gene (Walter et al, 2012). Both male and female can be affected. When both parents are carriers of the defective gene, with each pregnancy there is a 25% chance of the child inheriting the condition (1:4).

# **DIAGNOSIS:**

Babies with phenylketonuria are identified on the National Newborn Screening Card.

**National Newborn Bloodspot Screening Laboratory (NNBSL)**

Phenylketonuria is one of 6 conditions screened for on the National Newborn Screening card in Ireland. The sample is taken 72-120 hrs after birth. It is recommended to review the National Newborn Screening policy for guidelines regarding breast fed, preterm infants or infants that have not commenced feeding (National Newborn Screening Laboratory, 2011).

**Siblings of known cases of Phenylketonuria**

A serum sample is taken on Day 3 and Day 10 for plasma phenylalanine and tyrosine determination ([www.nnsp.ie](http://www.nnsp.ie)) and a newborn screening sample should be taken between 72 and 120 hours following birth (as for all other newborn infants).

**Confirmation**

When an elevated phenylalanine level is detected, the National Newborn Screening laboratory will request a serum sample to confirm the diagnosis of PKU (National Newborn Screening Laboratory, 2011).

# **THERAPEUTIC MANAGEMENT:**

Dietary treatment should commence as soon as the diagnosis is confirmed (Acosta, 2010). The baby is admitted to Temple Street Children’s University Hospital. The aims of the admission are:

1. To reduce phenylalaine level to therapeutic range

(120 – 360 umol/L <12yrs & 120-600umol/L >12yrs)

1. Support family following diagnosis
2. Provide initial education to parents/guardians
3. Instruct parents/guardians on blood sample collection
4. Introduction to the Multi-disciplinary team

**Levels**

Phenylalanine levels of greater than 360 micromoles / litre in a newborn infant necessitates commencement of a low protein diet. Scriver et al (2000) classify PKU as the following:

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| **Physiological levels of Phenylalanine in**  **non – PKU person** | 0 - 182 micromoles / L |
| **Classical PKU**  **Mild PKU**  **Non- PKU Hyperphenylalaninaemia (HPA)** | > 1200 micromoles / L    600 – 1200 micromoles / L  182 - 599 micromoles / L |
| **Therapeutic range in PKU Child** | 120-360 micromoles /l (allows for adequate growth) |

TSCUH Policy No. MU 014/01

The aims of dietary treatment are to;

1. Reduce phenylalanine levels to 120-360 µmol/l <12years & 120-600 µmol/l >12years
2. Provide adequate protein, energy, vitamins and minerals for growth and development.
3. Replace tyrosine and prevent deficiencies
4. Provide variety, palatability and flexibility to suit individual lifestyles.

When the Clinical Nurse Specialist meets with the parents initially she will offer advice regarding searching the internet for information on the condition. If any information found contradicts that given by the team, they are encouraged to discuss this to ensure there is clarity in the management of their child. Written information is given to the parents/guardians to take home to read for themselves and other family members. A DVD is also available to help educate other members of the family.

# **NURSING CARE OF NEWLY DIAGNOSED INFANT WITH CLASSICAL PHENYLKETONURIA**

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| ACTION | RATIONALE |
| **Refer to care plan for newly diagnosed PKU.**   1. **GENERAL OBSERVATIONS**  * Temperature / pulse / respiration& Blood Pressure   Record vital signs on admission and thereafter as dictated by patients condition.  **WEIGHT AND HEIGHT**  Monitor **weight and length** on admission. Further measurements as deemed necessary. | To provide baseline.  Vital signs are usually normal unless there is an underlying illness or infection.  Baseline Measurements |
| 1. **DIET**  **DIET IS FOR LIFE** Divided into **3** components  **1.** **Measured amounts of phenylalanine (natural protein).**    Babies (pre-weaning) will receive phenylalanine from their regular infant formula or breast milk.  **2. Synthetic Amino Acid mixture**  (phenylalanine-free infant formula / drink)  **3. Vitamins and minerals**    Avoid medications which have been sweetened using the substance **Aspartame eg. Some antibiotics, regular & diet drinks etc.**  Measured amount of natural protein and synthetic protein given with each feed as prescribed. These feeds should not be mixed together. | Phenylalanine is necessary in the diet to ensure adequate growth and development.  Natural protein intake varies from child to child and is titrated against blood phenylalanine levels.  Balances protein requirements. Volume of synthetic protein is based on total requirements for the infant’s age and weight.  There is potential for deficiency due to the diet restrictions. The synthetic protein supplement contains vitamins and minerals to prevent deficiencies.  Aspartame is a source of phenylalanine (Walker et al, 2003)  **It is important not to mix feeds so that:**  1. Exact amounts taken of each feed can be measured.  2. Taste of either feed remains unaltered. |
| 1. **EDUCATION**   **Teaching is a continuous process** and knowledge is evaluated at every opportunity.    **Diet**   * Liaise with dietetic team regarding teaching of diet. * Organise visit to the Formula Room for teaching and assessment of competency in making feeds.   **Blood letting**  Educate parents/guardians regarding the correct technique for obtaining blood sample from heel on blood spot card (teaching plan available).  Frequency of Blood samples  **Genetics**  The inheritance of PKU will be discussed with the parents/guardians by the metabolic team during the admission. | Initial teaching sessions will be targeted at teaching parents/guardians:  1. Rationale for diet  2. Difference between the two feeds i.e. synthetic and natural protein  3. Emphasize synthetic protein can be given to appetite. However, natural protein can only be given as directed.  It is important to ensure that at least one parent is competent in taking blood sample from the heel before discharge.  **On admission a sample is obtained for phenylalanine level and also for pterin levels.**  **\* *Please note, during admission, samples are taken on a card to demonstrate technique used at home. However, staff also need to send a daily tube sample in a Microvette tube (orange top) to lab for same day results.\****  Frequency of blood sampling will then continue as per medical direction and/or Metabolic unit policy MU 014/01  Ensure parents understand the 1:4 risk of inheritance of PKU in further pregnancies. |
| 1. **FOLLOW UP CARE / MULTIDISCIPLINARY TEAM**  * Metabolic Outpatients clinic for medical, dietetic and nursing support. * **Regular blood testing** for phenylalanine and tyrosine * **Psychology** * **Social Worker** | To provide on-going support, education and encouragement for the family. Frequency of OPD visits as per unit policy.  Support around diagnosis.  Introduction to service provided on an on-going basis.  Will ensure patient and family are aware of appropriate benefits and entitlements e.g. long-term illness card.  Will provide support around diagnosis |

# **REVIEW:**

This procedure shall be reviewed and updated at least every two years by the Clinical Education Facilitator, NCIMD in order to determine its effectiveness and appropriateness. It shall be assessed and amended as necessary during this period to reflect any changes in best practice, law, substantial organisational change and professional or academic change.

# **AUDIT AND EVALUATION**

In order to ensure the effectiveness of this policy and procedure the Clinical Education Facilitator NCIMD shall complete an audit annually to review and monitor compliance with this policy and procedure. The Clinical Education Facilitator NCIMD must further provide a systematic process for the reporting and investigation of compliance breaches, or potential breaches, to enable proactive prevention in the future.

# **REFERENCES:**

**Phenylketonuria**

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