TITLE: NURSING GUIDELINES FOR THE MANAGEMENT OF CHILDREN WITH GLYCOGEN STORAGE DISEASE TYPE 111

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1. 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.

2. DEFINITIONS:

Glycogen Storage Disease Type 3 (GSD III) is an autosomal recessive disorder and occurs as a result of a deficiency of glycogen debrancher enzyme (Crushell et al, 2010). As a result of the deficiency, the glycogen structure is altered and is stored in this manner (Laforet et el, 2012). Most patients with GSD III have an enzyme deficiency in the liver, muscle and heart (GSD IIIa) and a smaller amount (~ 15%) of patients only have liver symptoms (GSD IIIb) (Laforet et el, 2012). The enzyme defect allows only partial degradation of glycogen during fasting. Hypoglycaemia therefore occurs.

3. PRESENTATION:

The patient will present with hypoglycaemia (unable to fast for long periods of time).

Clinical features are as follows:

- Hypoglycaemia
- Lacticacidosis

- Moderate to severe elevation of liver function tests
- Hepatomegaly (due to glycogen and fat accumulation).
  - Protruding abdomen
  - Truncal obesity
- Hyperlipidaemia
- Short stature

(Kishnani et al, 2010: Laforet et el, 2012).

4. DIAGNOSIS:

- Diagnostic tests include a 24 - hour glucose / lactate profile, and a glucagon stimulation test (no increase in glucose levels as unable to release glycogen stores).
- The clinical findings are confirmed by:
  - red cell glycogen
  - leukocyte glycogen debrancher enzyme activity
  - AGL gene mutation analysis.
- All future new-borns within an affected family should have a "high risk" screen at birth.

5. MANAGEMENT:

Involves dietary management:

a) Ensure adequate carbohydrate intake to prevent hypoglycaemia.

b) Administer uncooked corn starch (UCS) / Glycosade (i.e. long-acting starch) throughout the day and night to prevent a decrease in glucose levels. Glycosade has been introduced as an alternative to UCS and has shown significant improvement in relation to quality of life (e.g. less disruption to sleep) (Bhattacharya et al, 2007; Correia et al, 2008). Infants and younger children are likely to require overnight feeding.

These therapeutic goals are even more important during periods of illness or stress.

Prompt recognition and early treatment will prevent coma, brain deterioration or even death.

6. COMPLICATIONS:

- Growth retardation.
- Dextran deposits in the liver and muscle due to partial degradation of glycogen acts as foreign body leading to cirrhosis and possible jaundice.
- Cardiomyopathy for type IIIa

- **Adult complications:**
  - Hepatic adenomas (rare)
  - Liver cirrhosis (rarely resulting in liver failure)

7. NURSING MANAGEMENT OF THE PATIENT WITH GLYCOGEN STORAGE DISEASE TYPE 111

Nursing observation and attention to detail is vital. The reporting of vomiting or diarrhoea may be lifesaving.

ACTION	RATIONALE
1. GENERAL OBSERVATIONS: **SKIN** Assess colour, pallor, temperature and clamminess of skin.	Patients with hypoglycaemia may present with these symptoms.
2. NEUROLOGICAL STATUS: Assess **neurological status** using a Glasgow **Coma Scale** if patient has altered level of consciousness, muscle weakness, lethargy and seizures. If ambulant, record and report any ataxia.	Due to hypoglycaemia, the brain is not receiving adequate amounts of energy (i.e. glucose) resulting in altered state of consciousness. The body draws on the protein as a source of energy, resulting in muscle wasting. This includes the heart muscle, resulting in cardiomyopathy.
3. VITAL SIGNS: Paediatric Early Warning System score will be used to monitor patient status (4 hourly – daily). ▪ **TEMPERATURE**	Full blood work-up required if pyrexial.

ACTION	RATIONALE
▪ **PULSE** ▪ **RESPIRATION**	Tachycardia may indicate infection or hypoglycaemia. Tachypnoea may indicate infection. Hypoglycaemia is not always associated with increased heart and respiratory rate as patients who have a diagnosis of GSD III can become accustomed to low glucose levels.
4. BLOOD GLUCOSE: • Recorded as per Glucose & Lactate Profile (PP-CLIN-NCIMD-14)	To ensure safety while correct feeding regime is being established, and to assess efficacy of dietary management.
5. URINALYSIS:	Ketones indicate fat catabolism.
6. DIET AND DIETARY EDUCATION: **The following diet is commenced;** • High carbohydrate intake. • Increased protein intake. • Low fat intake. Liaise with dietetic team in the education of parents and family.	 Prevention of hypoglycaemia. Counteract drain of protein from muscle and improve muscle function. Fat is metabolized in the liver. Parents must be aware of the risks and complications associated with poor compliance to diet.

ACTION	RATIONALE
7. FEEDING: Younger patients require more frequent feeding (i.e. 2-3 hourly and continuous enteral feeding at night). Use of enuresis mat. Having been monitored and deemed safe, the feeding intervals of older patients can be extended and enteral feeding may not be required at night. These patients may require a complex carbohydrate drink prior to sleep and a feed during the night. Ensure bottles are shaken well prior to feeding as cornflour / glycosade is the mainstay of treatment and is inclined to settle at the bottom of the bottle.	To prevent hypoglycaemia, induce catch-up growth, reduce liver size and decrease serum transaminases. Helpful indicator if child vomits or feed leaks as device will alarm. Glycosade and corn flour are slow releasing carbohydrates. Glycosade or corn flour are introduced to the diet from various ages and is consultant & dietetic guided. Cornflower should not be cooked or heated as heating disrupts the starch granules rendering the solution ineffective (Dixon, 1994).
8. MEDICATION: **Ibuprofen (when indicated)** **Paracetamol use is not advocated in**	Analgesia, antipyretic. Not metabolized in the liver. Metabolized in the liver (Higgins, 1996).

ACTION	RATIONALE
patients with GSD II	
9. INVESTIGATIONS: **Bloods** Cholesterol, Triglycerides, Creatine Phosphate-kinase(CK),Uric-Acid, Liver Function Tests (LFT's).	To detect and determine muscle and liver damage. Dietary revision may be required.
10. WEIGHT AND HEIGHT: Plot weight and height once a week while in hospital.	To assess growth and development. To ensure adequate CHO and protein is being given to meet needs.
11. MULTIDISCIPLINARY FOLLOW UP: • Metabolic Clinic for medical, dietetic and nursing support. • Some patients require 3-4 monthly admissions for 24 hour glucose monitoring and dietary assessment. • Blood tests as above	To monitor Liver Function and dietary compliance, growth and development.

ACTION	RATIONALE
• Psychology • Social Work • Radiology • Annual ECHO	To assess effects of long-term illness on family, difficulties maintaining diet and any other specific difficulties encountered. To ensure entitlements are received.

8. 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.

9. REFERENCES:

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