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

Revision: 1

Policy ☐ Procedure ☐ Protocol ☐ Guideline ☑

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CONTENTS

1. STATEMENT:

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 Inherited Metabolic Disorders (NCIMD).

Readers of this document are reminded that prescription of dietary regimes and all medications (including slow-release carbohydrates, 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. SCOPE

These guidelines are a point of reference for all nursing and medical staff in relation to the care of a child with Glycogen Storage Type III or suspected of having Glycogen Storage Disease Type III.

3. 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 al, 2012). Most patients with GSD III have an enzyme deficiency in the liver, muscle and heart (GSD IIIa) and a smaller number (~ 15%) of patients only have liver symptoms (GSD IIIb) (Sentner et al. 2016). The enzyme defect allows only partial degradation of glycogen during fasting. Hypoglycaemia therefore occurs.

4. PRESENTATION:

The patient will present with hypoglycaemia and will be unable to fast for long periods of time.

Clinical features are as follows:

- Hypoglycaemia
- Hyperlipidaemia
- Lactic acidosis
- Moderate to severe elevation of liver function tests including creatine kinase (CK)
- Hepatomegaly due to glycogen and fat accumulation.
    - Protruding abdomen
    - Truncal obesity
- Short stature
- Cardiomyopathy (Derks et al, 2015; Laforet et el, 2012).

5. DIAGNOSIS:

a. 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).

b. The clinical findings are confirmed by:
    a. red cell glycogen
    b. leukocyte glycogen debrancher enzyme activity
    c. AGL gene mutation analysis.

c. All future new-borns within an affected family should have a "high risk" screen at birth.

6. MANAGEMENT:

Involves dietary management:

a. Ensure adequate carbohydrate intake to prevent hypoglycaemia.

b. Offer and encourage uncooked corn starch (UCCS) / 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 UCCS 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 orally or via nasogastric or PEG tube.

These therapeutic goals are even more important during periods of illness or stress. Patient may have to commence intravenous fluids containing glucose during extended periods of fasting or due to vomiting or diarrhoea to maintain normoglycemia.

7. 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)

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

Complete full nursing assessment on admission and continue to observe for signs of patient deterioration such as increased vomiting, diarrhoea and altered Glasgow Coma Scale score (GCS). Early detection of symptoms with prompt escalation of care and treatment may be lifesaving (Brown et al. 2015).

ACTION	RATIONALE
8.1. Emergency Assessment Complete patient assessment on admission and document vital signs in Paediatric Early Warning System (PEWS) chart. Escalate care as indicated by clinical judgment and PEWS score. Frequency of monitoring will be dictated by patient's condition (i.e. 2-4 hourly during initial presentation and acute illnesses - Paediatric Early Warning Score (PEWS) is used in CHITS.)	
Airway	Airway patency may be compromised due to altered Glasgow Coma Scale. Due to hypoglycaemia, the brain does not receive adequate amounts of energy (i.e. glucose) resulting in altered state of consciousness. Seizure activity due to hypoglycaemia may also compromise airway patency.

ACTION	RATIONALE
Breathing Monitor respiratory rate, respiratory effort and oxygen requirements as per PEWS. Obtain oxygen saturation levels if concerned and report abnormalities to medical team.	Tachypnoea, increased respiratory effort, reduced oxygen levels and increased CO_2 levels may indicate; • Infection • Underlying Respiratory Illness • Hypoglycaemia • Cardiomyopathy Patients with GSDIIIa may have cardiac involvement and cardiomyopathy, generally presenting in childhood (Sentner et al. 2016). Some children with cardiomyopathy may present with a feeling of incomplete inhalation or shortness of breath, particularly during exercise (Tril et al. 2019).
Circulation ➢ **Pulse**	Tachycardia may occur as a result of • Infection • Fluid overload • Dehydration • Hypoglycaemia • Cardiomyopathy Patients with cardiomyopathy may also present with bradycardia or an irregular heart rhythm (Tril et al. 2019).

ACTION	RATIONALE
➢ **Include Capillary Refill Time (CRT) assessment as per PEWS**	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.
➢ **Blood pressure** Report abnormalities in vital signs to the metabolic team.	Can be hypotensive with hypovolemic shock or dehydration. Beta blockers and nondihydropridine calcium channel blockers are the mainstay of therapy in symptomatic cardiomyopathy (Cassagnol, 2016). Patients with cardiomyopathy may be hypotensive as a result of these medications.
Disability Assess and record baseline neurological status using **Glasgow Coma Scale**, and continue to record especially during further episodes of acute illness. Report any abnormalities or deterioration to the Metabolic Team.	Due to hypoglycaemia, the brain does not receive adequate amounts of energy (i.e. glucose) resulting in altered state of consciousness. The body draws on the protein from its own muscle stores as a source of energy, resulting in muscle wasting. This includes the heart muscle resulting in cardiomyopathy.

ACTION	RATIONALE
➢ Observe and report muscle weakness and seizure activity. Record seizure type, duration and intervention and record seizure activity in nursing notes. Report any abnormal patient movements to the medical team. If ambulant, record and report any ataxia.	Seizures resulting from hypoglycaemia and encephalopathy.
Exposure to ensure full examination (whilst respecting the child's dignity and ensuring body temperature conservation).	
➢ **Temperature** Monitor temperature 4 hourly or more frequently if indicated.	Pyrexia may indicate presence of infection. Consider in conjunction with other signs such as tachycardia and delayed central capillary refill time. Follow Sepsis 6 protocol in PEWS chart. The following should be performed, blood cultures, F.B.C., U+E, LFTs blood gas and urine for culture and sensitivity. Hypothermia may indicate overwhelming infection (Goldstein et al. 2005).
➢ **Skin** Assess colour, pallor, temperature and clamminess of skin.	Patients with hypoglycaemia may present with these symptoms.

ACTION	RATIONALE
8.2. BLOOD GLUCOSE: Recorded as per Glucose & Lactate Profile (PP-CLIN-NCIMD-14). Monitor blood ketones.	Baseline assessment. To monitor for signs of hypoglycaemia and dehydration so that early treatment can be implemented. To assess efficacy of dietary management. Monitor more frequently if alterations made to diet as recommended by metabolic consultant.
8.3. INVESTIGATIONS: **BLOODS:** Cholesterol, Triglycerides, Creatine Kinase (CK), Uric-Acid, Liver Function Tests (LFTs), Ketones. **URINE OR BLOOD KETONES:**	To detect and determine muscle and liver abnormalities Dietary revision may be required. Ketones indicate fat catabolism.
8.4. DIET AND DIETARY EDUCATION: **The following diet is commenced;** • High carbohydrate intake. • Increased protein intake.	 Prevention of hypoglycaemia. Counteract drain of protein from muscle and improve muscle function. Gluconeogenesis may be increased due to high protein supply (Scholl-Burgi et al. 2015).

ACTION	RATIONALE
Liaise with dietetic team in the education of parents and family.	Parents must be aware of the risks and complications associated with poor compliance to diet.
8.5. FEEDING: Younger patients require more frequent feeding (i.e. 2-3 hourly and continuous enteral feeding at night). Use an 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 corn flour / glycosade is the mainstay of treatment and is inclined to settle at the bottom of the bottle.	To prevent hypoglycaemia, enable 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 & dietician guided. Cornflower should not be cooked or heated as heating disrupts the starch granules rendering the solution ineffective (Kishnani et al. 2014).

ACTION	RATIONALE
8.6. MEDICATION: **Ibuprofen (when indicated)** **Paracetamol use is not advocated in patients with GSD III.**	Analgesia, antipyretic. Ibuprofen not metabolised in the liver. Paracetamol is metabolised in the liver (Malar and Bai, 2012).
8.7 WEIGHT AND HEIGHT: Plot weight and height on age appropriate growth chart once a week while in hospital.	To assess growth and development. To ensure adequate CHO and protein is being given to meet needs.

8.8. MULTIDISCIPLINARY FOLLOW UP:	
• Metabolic Clinic for medical, dietetic and nursing support.	
• Some patients require admissions every 3-4 months for 24-hour glucose monitoring and dietary assessment.	To monitor Liver Function and dietary compliance, growth and development.
• Blood tests as above	
• Psychology	To assess effects of long-term illness on family, difficulties maintaining diet and any other specific difficulties encountered.
• Social Work	To ensure entitlements are received.
• Radiology	
• Annual ECHO	Due to risk of cardiomyopathy in GSDIIIa (Sentner et al. 2016).
• Annual abdominal ultrasound	To monitor as at risk of hepatic adenomas (Sentner et al. 2016).

9. MONITORING, AUDIT & EVALUATION

This procedure shall be reviewed and updated at least every three 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.

10. KEY STAKEHOLDERS

The following Key Stakeholders were consulted in the development/review of this document:

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