Acute management of patients 0 -18 years presenting with diagnosed metabolic conditions

UHL Children's Medical Guideline

University Hospitals of Leicester NHS
NHS Trust

Leicester Children's Hospital

Trust ref: E3/2020

1. Introduction and who this guideline applies to:

Current National Guidelines for the acute management of patients (0-18 years) presenting with diagnosed metabolic conditions should be accessed electronically via the British Inherited Metabolic Disorders Group (BIMDG) website (hyperlink below). Parents/carers may have a handheld copy of the appropriate medical guideline.

https://bimdg.org.uk/guidelines/

This guideline is intended for the clinical staff caring for patients presenting with diagnosed metabolic conditions.

Please liaise with the Children's Metabolic Team regarding the management of metabolic patients for elective surgery.

Related Documents:

Paediatric Metabolic Emergencies CoMET Guideline Trust Ref B12/2023

Medium Chain Acyl Co A Dehydrogenase Deficiency (MCADD) Paeditric UHL Nutrition and Dietetics Guideline Trust Ref C30/2016

Medium Chain Acyl CoA Dehydrogenase Deficiency (MCADD) - Newborn Infants UHL Neonatal Guideline Trust ref: C4/2012

Guidelines for the management of undiagnosed hyperammonaemia can be sourced via the BIMDG hyperlink provided, for UHL Intensive care patients please also see Hyperammonaemia UHL Childrens Intensive Care Guideline Trust ref: C50/2016

2. Acute management of patients (0-18 years) presenting with diagnosed metabolic conditions

Children with the metabolic disorders listed below must be prioritised and assessed urgently as they are at high risk of serious complications. An alert must be placed on NerveCentre if not already present.

- ***Citrin Deficiency***

- ***Fructose-1,6-Bisphosphatase deficiency***
- ***GLUT 1 deficiency***
- ***Glutaric Aciduria Type 1 (GA1)***
- ***Glycogen Storage Disorders (GSD) types 1,1b,3.***
- ***HMG CoA Lyase deficiency***
- ***Isovaleric acidaemia (IVA)***
- ***Ketotic Hypoglycaemia-****also covers glycerol kinase deficiency and glycogen synthase deficiency, and should also be used for GSD 9.*
- ***Long chain fat oxidation disorders -*** covers VLCAD, LCHAD, CPT II (severe early onset variants) carnitine translocase (late onset). Multiple acyl CoA dehydrogenase deficiency (MADD)
- ***Maple syrup urine disease (MSUD)***
- ***Medium Chain Acyl CoA Dehydrogenase deficiency (MCADD)***
- ***Methylmalonic Acidaemia (MMA)***
- ***Propionic Acidaemia (PA)***
- ***Urea cycle disorders 1 -*** covers OTC (OCT, ornithine transcarbamylase, ornithine carbamyl transferase) deficiency and CPS (carbamyl phosphate synthetase) deficiency.
- **Urea Cycle Disorders 2 -** covers citrullinaemia (Argininosuccinic synthase (ASS)) deficiency and argininosuccinic aciduria (argininosuccinic lyase (ASL)) deficiency.

Metabolic Team:

Consultant Paediatrician – ext 17933
Children's Metabolic Specialist Nurse – ext 17854
Specialist Dietitian – ext 175400

On call Metabolic Consultant (24 hour advice) – on mobile via Royal Manchester Children's Hospital switchboard 0161 2761234

3. Education and Training

None.

4. Monitoring Compliance

What will be measured to monitor compliance	How will compliance be monitored	Monitoring Lead	Frequency	Reporting arrangements
Compliance with national guidance for children with metabolic conditions	Audit of patient notes/datix	Paediatric Metabolic disorders Consultant	Annual	Local audit committee
Delays in the urgent assessment of children with metabolic conditions listed in Section 2	Datix	Paediatric Metabolic disorders Consultant	Annual	Local audit committee
Missing alerts on NerveCentre for patients with known metabolic condition	Datix	Paediatric Metabolic disorders Consultant	Annual	Local audit committee
Any difficulties in accessing National Guidelines electronically	Datix	Paediatric Metabolic disorders Consultant	Annual	Local audit committee

5. Supporting References

British Inherited Metabolic Disorders Group
(http://www.bimdg.org.uk/site/guidelines.asp accessed April 2024)

6. Key Words

Citrin Deficiency, Fructose-1,6-Bisphosphatase deficiency, GLUT 1 deficiency, Glutaric Aciduria Type 1 (GA1), Glycogen Storage Disorders (GSD) types 1,1b,3., HMG CoA Lyase deficiency, Isovaleric acidaemia (IVA), Ketotic Hypoglycaemia, Long chain fat oxidation disorders, Maple syrup urine disease (MSUD) , Medium Chain Acyl CoA Dehydrogenase deficiency (MCADD), Methylmalonic Acidaemia (MMA), Propionic Acidaemia (PA), Urea cycle disorders 1, Urea Cycle Disorders 2

The Trust recognises the diversity of the local community it serves. Our aim therefore is to provide a safe environment free from discrimination and treat all individuals fairly with dignity and appropriately according to their needs.
As part of its development, this policy and its impact on equality have been reviewed and no detriment was identified.

CONTACT AND REVIEW DETAILS	
Guideline Lead (Name and Title) J Forster – Consultant	**Executive Lead** **Chief Medical Officer**
Details of Changes made during review: **April 2024** Updated contact numbers. Updated related documents Added NerveCentre alert notification Added Trust equality statement Reviewed and updated Monitoring Compliance	