P427 KETOTIC HYPOGLYCAEMIA: AN AUDIT OF ADHERENCE TO LOCAL GUIDELINES

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Background Ketotic Hypoglycaemia (Blood sugar <3 mmol/L and blood ketones >2.5) is common in children with acute gastroenteritis and other common illnesses with poor oral intake. Untreated hypoglycaemia can lead to serious neurological sequelae.

Some metabolic disorders can present with ketotic hypoglycaemia, the presenting illness unmasking the underlying metabolic problem, despite the presence of ketones. We looked specifically at cases of ketotic hypoglycaemia in children with a history of diarrhoea and vomiting or acute illness with inadequate oral intake.

Aims To compare screening and management of ketotic hypoglycaemia in association with acute illness with the local guidelines.

Methods Patients were identified through HIPE code "E-16-2".

A retrospective chart review was done on patients presenting with hypoglycaemia from Feb 2016 to May 2017 to compare the screening and management of patients with the local guidelines.

As per audit recommendation the local hospital guideline was reviewed and an Algorithm along with "Hypoglycaemia pack" was made available in the acute assessment unit.

Staff education session was arranged A re-audit was performed on patients presenting with hypoglycaemia from January 2018 to December 2018 to see the adherence to the recommendations.

Results The following table shows comparison of initial audit and re-audit results

Abstract P427 Table 1

Year	2016/17 Audit	2018 Re-audit
Total Patients (n)	14	12
Male	5	4
Female	9	8
Median age	18 Months	36 months
Mean Age	21.5 months	51 months
Lab glucose checked	7/14 (50%)	11 (91.6%)
Serum AA checked	10/14(71.4%)	9/12 (75%)
UrinaryOA checked < 24 hours	4/14 (33.3%)	12/12(100%)
Time taken to recheck blood glucose after initial management	Median 42.5 minutes Mean 64 minutes	32.5 minutes 37.5 minutes

Conclusion Re audit showed marked improvement in lab glucose monitoring ,rechecking blood glucose timing and taking samples for urinary organic acids.However there is still room for improvement in sampling serum Amino acids.

All patients got treatment as per guidelines.

Recommendations Algorithmic approach,staff education and availability of "Hypoglycaemia Packs" did improve the outcome however there is continued need for staff education to adhere to the guidelines

P428 AN INTERESTING CASE OF A GIRL WITH A DE NOVO UNBALANCED TRANSLOCATION CAUSING DISTINCTIVE METABOLIC MARKERS

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Background A five year-old girl was referred to the National Centre for Inherited Metabolic Disorders with a background of a neonatal seizure on day three of life in the context of hypoglycaemia and weight loss. She had learning difficulties and gross motor and speech delay. To investigate her developmental delay an MRI brain was carried out at 3 years of age which demonstrated extensive signal abnormality in the deep white matter with numerous tiny areas of high T2 and FLAIR signal abnormality which was felt reflected either neonatal hypoxic ischaemic encephalopathy or neonatal hypoglycaemia or both. A metabolic cause was considered at that time and an initial screen was normal; the patient had also undergone a microarray.

Investigations Metabolic investigations were conducted in parallel with detailed genetic studies to search for an encompassing diagnosis. The girl was diagnosed with a *de novo* unbalanced translocation between the long arm of chromosome X and duplication of the long arm of chromosome 13, der (X)t(X;13)(q25;q12.11)dn.

A blood sample revealed slightly abnormal glycosylation of transferrin isoforms with increased loss of one sialic acid (8.3%, reference range <6%). This was repeated with a result of 8.6%. However it was felt that this was not consistent with a primary diagnosis of congenital disorder of glycosylation (CDG) type 1 and not definitive of a CDG type 2.

Results On review of all of the results it was felt that the genetic abnormality identified was responsible for this girl's clinical presentation. Her MRI seemed more likely to reflect a developmental brain disorder or reflective of the neonatal events. There are genes for CDG on both the X chromosome and chromosome 13 but neither appear to be at the position of her breakpoint; this could explain her distinctive glycosylation abnormalities as a secondary phenomenon.

Conclusions Our patient is clinically making pleasing developmental progress though recently had a recurrence of seizures after a prolonged seizure free period. This case demonstrates how important it is to consider all the biochemical and genetic information available in a clinical context to differentiate between primary and secondary findings. This will guide towards accurate diagnostic labelling, streamline follow-up and enable appropriately directed care.