

years was on the 25<sup>th</sup> centile and it had been steadily increasing over 18 months. He was not dysmorphic and had a mild degree of hirsutism.

**Results** The investigations did not reveal dyslipidaemia or evidence of hyperglycaemia. Thyroid functions and insulin like growth factor-1 were within normal limits. His alanine transaminase level was mildly elevated (70 IU/L). An ultrasound scan revealed fatty liver.

A SNP array was performed and a 604kb interstitial heterozygous deletion of 16p11.2 chromosome from base pair 29,595,483 to 30,199,713 was identified. The detected deletion is in the recurrent proximal 16p11.2 deletion syndrome region, and deletion syndromes in this region have been associated with autism, developmental delay, and obesity; however, our literature search did not reveal cases with fatty liver presenting at this age.

#### Conclusion

**Discussion** This case highlights the importance of considering genetic investigations in children with rapid weight gain, wherein nutritional factors are not obviously contributing to weight gain. The fatty liver will need on-going follow-up.

### 1325 THYROID FUCTION TESTS IN PALPITATIONS

Pramod Nair, Tabrez Noorani, Rajesh Sesham, Olamide Shekoni. *Bedford Hospital NHS Trust, Bedfordshire Hospitals*

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**Aims** To determine if thyroid function tests are useful in the investigation process of a child presenting with palpitations

**Methods** We looked at 87 children presenting with palpitations to the paediatric outpatient clinic over a 2 year period. We selected the children who had thyroid function tests done as a part of their investigations. We excluded children with known thyroid problems either hypo or hyperthyroidism

**Results** Of the 87 children, there were 52 females and 35 males. The median age was 13 years with patients ages ranging between 6-17 years. The presenting complaint was palpitations and all of them had blood tests including thyroid function tests either done by GP or the paediatrician. None of the patients had a note of goitre or other symptoms of hyperthyroidism. Of the 87 patients, 75 had completely normal thyroid values. 12 patients had a high TSH ranging between 4.3-6.4. All 12 patients were reviewed and followed up with thyroid functions normalizing or considered to be within acceptable ranges. None of the patients had tests suggestive of hyperthyroidism.

**Conclusion** Hyperthyroidism is known to cause palpitations in children. Although thyroid function tests are requested routinely as part of investigations of children with palpitations either in primary care or hospital they have a poor yield in terms of diagnosis of hyperthyroidism. Occasionally the thyroid function tests might show slightly deranged levels of TSH which might then worry their parents and necessitate further investigations. Given this study, we feel routine use of thyroid function tests is unnecessary in a child presenting with palpitations and unless there are other clinical features of hyperthyroidism then these tests should not be undertaken.

### 1331 ROLE OF HYPERPARATHYROIDISM AND VITAMIN-D DEFICIENCY IN PRETERM INFANTS WITH METABOLIC BONE DISEASE

Barah Hassan, Prakash Kannan Loganathan. *South Tees Hospital NHS Foundation Trust*

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**Aims** Metabolic bone disease of prematurity (MBD) is relatively common in preterm and low birth weight infants and can be associated with significant morbidity. Screening and diagnosis of this condition includes serial investigations for calcium, phosphate, and alkaline Phosphatase (ALP). Commonly, oral phosphate (Po4) supplements were prescribed presuming Po4 deficiency/loss as the underlying primary pathology. Role of serum parathyroid hormone (PTH) and vitamin-D levels in diagnosis/management of MBD is understudied.

This study aims to identify the prevalence of secondary hyperparathyroidism and vitamin-D deficiency in neonates at high risk and/or diagnosis of metabolic bone disease of prematurity.

**Methods** In this single center study, preterm infants who had either a PTH or 25 hydroxyvitamin-D performed between the years 2017-2021 were identified from the hospital pathology system. Laboratory details were collected from hospital pathology system (webICE) and demographic details from neonatal database (Badger). Concurrent serum calcium, phosphate and ALP measurements were also recorded. We defined serum PTH (>7pmol/L) as hyperparathyroidism, 25-hydroxyvitamin D <50 nmol/L as inadequate Vitamin D level, hypophosphataemia as <1.8mmol/L, hypocalcaemia as <2mmol/L and raised ALP as >500u/L. This audit was registered with hospital audit department.

**Results** A total of 430 patients between 2017-2021 were admitted to the neonatal unit with birth weight below 1.5 kilograms and/or gestation below 32 weeks. 35 (8%) babies were identified as having either a PTH and/or vitamin D level performed. All 35 babies were between 24 – 28 weeks' gestation at birth and 29 (83%) of the babies were born below 1 kilogram. Mean gestational age was 25.5 ± standard deviation (SD) 1.29 weeks and birth weight 766 ± 210 gra table 1 outlines the median laboratory results.

Initial samples were collected at variable timings. For PTH, 5 (15%) were before 20 days of life, 11 (33%) between 20-39 days, 5 (15%) between 40-59 days and 12(36%) beyond day 60. For vitamin-D, 1 (5%) was before day 20 of life, 2 (10%) between 20-39 days, 7 (35%) between 40-59 days and 10 (50%) beyond day 60.

Table 2 outlines the numbers of patients with abnormal results. Of the twenty-three patients who had a raised PTH, 5 (22%) had normal biochemistry bloods. For those who had vitamin-D levels measured, 2 (10%) of the 20 patients had normal biochemistry bloods – both had normal vitamin-D levels but raised PTH. Raised ALP and low phosphate was reported in 63% and 57% of audited patients respectively – highlighting these as the most common abnormalities in screening bloods. Hypocalcaemia appeared to be relatively rare with only 9% of reported cases

**Abstract 1331 Table 1** Median laboratory results with inter-quartile range (IQR) for the first testing

| PTH level (pmol/L), n = 33       | 25-hydroxyvitamin D level (nmol/L), n = 20 | Ca level (mmol/L), n = 36      | PO4 level (mmol/L), n = 30     | ALP level (u/L), n = 35     |
|----------------------------------|--|--------------------------------|--------------------------------|-----------------------------|
| Median (IQR): 16.65 (6.9 – 26.4) | Median (IQR): 53.4 (42.5-64.1)             | Median (IQR): 2.51 (2.43-2.59) | Median (IQR): 1.75 (1.57-1.98) | Median (IQR): 585 (471-120) |

**Abstract 1331 Table 2** Numbers of patients with abnormal blood results

| Laboratory blood result (sample size)              | Number of patients (percentage %) |
|--|-----------------------------------|
| Hyperparathyroidism: serum PTH (>7pmol/L) (n = 31) | 23 (74%)                          |
| 25-hydroxyvitamin D <50 nmol/L (n = 20)            | 10 (50%)                          |
| Hypocalcaemia: Serum calcium <2mmol/L (n = 35)     | 3 (9%)                            |
| Hypophosphatemia: Serum Po4 <1.8mmol/L (n = 30)    | 17 (57%)                          |
| Raised ALP >500u/L (n = 35)                        | 22 (63%)                          |

**Conclusion** Very few preterm infants underwent PTH and/or a vitamin-D analysis, and investigated at varying ages, as part of MBD management. Significant proportion of preterm infants had secondary hyperparathyroidism (74%) and inadequate vitamin D levels (50%). These infants would benefit from oral calcium and vitamin D supplementation rather than phosphate supplementation. Further research is needed to assess the usefulness of serum PTH, vitamin D levels and role calcium supplementation in the management of MBD.

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**TYPE 1 DIABETES MELLITUS: EARLY DIAGNOSIS AND REFERRAL**

<sup>1</sup>Irfan Nengroo, <sup>2</sup>Kumudu Pematilleke, <sup>3</sup>Tanya Naydeva. <sup>1</sup>ST5; <sup>2</sup>Paediatric Registrar at Lincoln County Hospital; <sup>3</sup>Consultant Paediatrics with special interest in Diabetes at Lincoln County Hospital

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**Aims** Around 3000 new Paediatric Diagnosis of Type 1 Diabetes Mellitus a year in the UK, with around 25% presenting in Diabetic Ketoacidosis. Prompt recognition and referral of children with suspected Diabetes is vital to prevent life threatening Diabetes Ketoacidosis and its consequences. NICE recommendations state to refer children and young people with suspected Type 1 Diabetes Mellitus immediately (on the same day) to a multidisciplinary Paediatric Diabetes team.

Our Aims were to:

Establish the current practice in diagnosing Type 1 Diabetes in children at a District General Hospital.-Compare the current practice to a set standard of practice by NICE and BSPED (see table 1 (Diabetes in children and young people Quality standard [QS125] Published: 14 July 2016)).

-Identify potential areas of improvements and concerns.

-Give recommendations based on the results of the study.

**Methods**

**Population** Patients ages 16 and below who were admitted to Lincoln County Hospital and Pilgrim Hospital Boston with a new Diagnosis of Type 1 Diabetes.

Retrospective study from 01/03/2020 till 31/12/2020 with data collection done from November 2020 till January 2021. Information extracted from Patients medical notes and Questionnaire see figure 1 (Questionnaire: Early Diagnosis of Type 1 Diabetes Audit 2020, Lincoln County Hospital).

**Results** Total patients included 25 (22 at Lincoln, 3 at Boston)

Age groups- 0-5 years : 2 patients 6-10 years: 6 patients, 11-16 years : 14 patients.

sex- Females 8, Males 17.

43% Presented with Diabetic Ketoacidosis and 57% were not in Diabetic Ketoacidosis.

Diagnosis of Type 1 Diabetes and Diabetic Ketoacidosis was done as per the NICE/BSPED clinical guidelines.

-Duration of symptoms before presenting to GP or A&E were between 2 days and 6 weeks.

-Delay in review by GP in 12%.-

Delay in referral to Paediatric team in 8%.

**Reasons for delay were** -One patient with glycosuria was treated for UTI and had a delay in referral to the Paediatric team of 2 weeks.

-Awaiting negative Covid screen results, average delay of 3 days till GP review.

- GP arranged bloods as Out Patient, with no referral to Paediatric team on the same day.

**Early Diagnosis of Type 1 Diabetes Audit Questionnaire**

1) What was the initial presentation of Type 1 Diabetes? Please tick the below options:

- hyperglycaemia
- polyuria
- polydipsia
- weight loss
- excessive tiredness
- Other- Please specify

2) How long did they have these symptoms for before the diagnosis of Type 1 Diabetes?

Please specify duration: \_\_\_\_\_

3) How many health care professionals did you see with these symptoms before getting the diagnosis of Type 1 Diabetes?

Mention number: \_\_\_\_\_

4) Were they referred to the Paediatric team on the same day for suspicion of Diabetes/ diagnosis of diabetes?

- Yes
- No

5) If you answered no to the above question, please mention the duration of delay?

\_\_\_\_\_

6) What was the reason for the delay?

\_\_\_\_\_

7) Were they seen immediately after being referred?

- Yes
- No

8) If you answered no to the above question, please mention the duration of delay?

\_\_\_\_\_

9) What was the reason for the delay?

\_\_\_\_\_

**Abstract 1316 Figure 1****Abstract 1316 Table 1**

| Clinical standards  | Target |
|---|--------|
| Immediate ( same day) referral of suspected Type 1 Diabetes to multidisciplinary paediatric diabetic team | 100%   |
| Measurement of blood glucose, ketone, pH in suspected DKA cases on arrival to hospital                    | 100%   |
| Immediate assessment of DKA cases for suspected cerebral oedema   | 100%   |

**Conclusion** Diagnosis was delayed by >24 h (median 3.0 days, range 1-14 days) in 20% (8)

Our recommendations