

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

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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

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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