low doses and robust PTx function. Home blood glucoses (BG) were controlled and HbA1c stable at 6% off insulin therapy. Eighteen months later, he was hospitalized for hyperglycemia with ketosis, without acidosis, during an upper respiratory infection. He was restarted on insulin (glargine, aspart). Once the acute stress resolved, his insulin dose needs reduced significantly, tapering down to insulin glargine 5 units daily. Along with daily home BG monitoring, HbA1c and C-peptide are performed every 3 months, with recent HbA1c 5.2% and C-peptide 3.0 ng/ml (simultaneous PG of 103 mg/dl).

Discussion

Causes of post-PTx hyperglycemia include graft failure due to acute or chronic rejection, insulin resistance, beta cell dysfunction due to IS medications, pancreatitis, or, rarely, recurrence of autoimmunity. Factors predicting hyperglycemia are pre-PTx insulin dose, BMI and acute rejection. New onset type 2 diabetes can occur due to insulin resistance from IS medications and genetic predisposition. Measurement of C-peptide after OGTT can help determine the cause. Insulin is the standard treatment even with detectable C-peptide, though oral glucose-lowering medications have been used in the setting of insulin resistance. In this patient, severe hyperglycemia occurred during stress. He had very low insulin dose requirements when the stress and hyperglycemia resolved and reasonable C-peptide values. This scenario was most consistent with hyperglycemia due to insulin resistance and subsequent relative insulin deficiency during stress, with continued PTx function. This illustrates the importance of detailed assessment and personalized treatment in patients with post-PTx hyperglycemia.

Adrenal

ADRENAL CASE REPORTS II

17-beta Hydroxysteroid Dehydrogenase 3 Deficiency in 1 Month Old Infant

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SUN-159

Background: 17-beta hydroxysteroid dehydrogenase 3 deficiency is a rare autosomal recessive disorder caused by mutations in HSD17B3 encoding the enzyme which converts androstenedione to testosterone. It is characterized in 46, XY males by incomplete virilization, including micropenis and hypospadias.

Clinical Case: We report a 1 month old infant who presented with ambiguous genitalia. Prenatal non-invasive screening showed a Y chromosome, however, fetal ultrasound revealed female genitalia. The infant was born with micropenis (~1.4 cm in length) and proximal hypospadias, with enlarged labioscrotal folds and palpable gonads bilaterally. The urethral meatus had been relocated surgically to the glans. There was an apparent vaginal orifice with a normally positioned anus. Initial testing revealed a normal serum 17-OHP (90 ng/dl, n<200 ng/dl) and normal electrolytes. Abdominal US showed normal kidneys. Pelvic US demonstrated no Mullerian structures; gonads thought to be testes were identified in the labioscrotal folds. At 3 months of age, the infant underwent a 3 day HCG stimulation testing with a borderline testosterone response to 132 ng/dl, androstenedione 78 ng/dl and DHT 25 ng/dl. T/A ratio was unremarkable at 1.7 (n>0.8). Thus, hormonal testing was unsupportive of a testicular steroidogenic enzyme deficiency or androgen insensitivity syndrome. Karyotype was confirmed as 46, XY with microarray evidence of multiple regions of homozygosity. Genotyping with a 46, XY DSD panel (GeneDx) revealed a homozygous pathogenic variant c.608 C>T (p.A203V) in exon 9 of the HSD17B3 gene, consistent with a diagnosis of autosomal recessive 17-beta hydroxysteroid dehydrogenase 3 deficiency. Parents are of Arabic descent and are consanguineous. An older brother was also born with ambiguous genital and was later found to be homozygous for the same mutation. This mutation has been identified in the homozygous state in several unrelated affected patients. Previously published functional studies demonstrated loss of enzymatic activity with this missense mutation (1). Male gender was assigned at birth, and parents wish to continue male sex of rearing.

Conclusion: Molecular genetic analysis utilizing a commercially available candidate gene panel for 46, XY disorders of sex development diagnosed 17 beta-HSD3 deficiency in this case where hormonal testing was not informative. Early and correct diagnosis is key in planning medical treatment to facilitate pubertal development.

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Bone and Mineral Metabolism

OSTEOPOROSIS: DIAGNOSIS AND CLINICAL ASPECTS

Hyponatremia Correction Normalizes Bone Mineral Density and Bone Fragility in Rats.

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SUN-371

Multiple epidemiologic studies have associated chronic hyponatremia with both osteoporosis and bone fractures. Studies in experimental animals and cultured cells have demonstrated that reducing the extracellular sodium concentration ([Na+]i) causes bone loss primarily by increasing osteoclast formation and bone resorbing activity. In osteoclastic cell cultures, reducing [Na+]i activated biochemical and functional changes in osteoclast activity, which appear to occur by direct sodium-sensing mechanisms on osteoclasts that are independent from changes in osmolality. Whether the pathological changes in bones induced by hyponatremia can be reversed by correction of hyponatremia has not been studied. The present studies were initiated to address this question. 22-month-old F344BN F1-hybrid rats were made hyponatremic using a desmopressin continuous infusion while fed a liquid diet. After 3 months of chronic sustained hyponatremia, a cohort of the hyponatremic rats were corrected to a normal [Na+]i by removal of the desmopressin minipumps.
and allowed to recover for 2 months. Both bone density measurements by DXA and biomechanical testing were performed on excised bones from normonatremic control rats (NN, [Na⁺]=145±5.6 mmol/L, n=9), chronically hyponatremic rats (HN, [Na⁺]=114±4.7 mmol/L, n=9) and hyponatremic corrected rats (HC, [Na⁺]=139±2.1 mmol/L, n=9). The results confirm that chronic hyponatremia caused significant decreases in bone mineral density (BMD, g/cm²) in the L4 vertebra (NN=0.166±0.003, HN=0.151±0.002, P=0.002) and femur (NN=0.229±0.004, HN=0.213±0.005, P=0.024). Bone fragility as measured by ultimate load to fracture (UL, Newtons) was also increased in the L5 vertebra (NN=369.8±51.1, HN=262.9±29.4, P=0.0001), but only slightly in the femur (NN=295.4±57.5, HN=286.0±35.4, P=0.682). Following correction of hyponatremia, both BMD and UL recovered after 2 months to levels not significantly different than the normonatremic controls: L4 BMD (NN=0.166±0.003, HN=0.167±0.002, NS); femur bone density (NN=0.229±0.004, HN=0.226±0.003, NS); UL in the L5 vertebra (NN=369.8±51.1, HN=262.9±29.4, P=0.0001), but only slightly in the femur (NN=295.4±57.5, HN=286.0±35.4, P=0.682). These results show that bone fragility parameters are adversely affected by chronic hyponatremia in addition to the previously reported decreases in BMD in rats. Consistent with the BMD results, the trabecular bone in the spine was more severely affected than the cortical bone in the femur. Our results show that much of the bone pathology of hyponatremia-induced osteoporosis can be reversed following correction of the hyponatremia without using specific antiresorptive therapy. Our findings therefore raise the possibility that correction of hyponatremia may be effective as a therapy for treatment of hyponatremia-induced osteoporosis in selected patients.

Pediatric Endocrinology

PEDIATRIC ENDOCRINE CASE REPORTS I

Is There an Association Between the Detection Method for Pediatric Thyroid Nodules and the Risk of Malignancy?

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SAT-079

Background

Thyroid nodules are less common in children compared to adults, but have a higher likelihood of malignancy. There are few studies, particularly in the pediatric population, examining the association between how and by whom the nodule is detected and the risk of malignancy. Several adult studies have suggested a high rate of malignancy in incidentally discovered thyroid nodules (1,2). However, this was not similarly seen in pediatric thyroid nodules according to one study (3). As fine needle aspiration (FNA) in pediatric patients may be more labor intensive and diagnostic excision is the recommendation for nodules with indeterminate or potentially malignant cytology, if the detection method can be a predictive measure of malignancy, it may enhance the evaluation of pediatric thyroid nodules.

Objective

The aim of this study is to determine if there is an association between how and by whom a thyroid nodule is detected and the risk of malignancy in the pediatric population.

Study Design and Methodology

We retrospectively reviewed the medical records of pediatric patients (≤21 years of age) who had a thyroid nodule with definitive cytologic or pathologic diagnosis from January 2010 to June 2019. Patients were categorized into 3 groups based on how and by whom the nodule was detected: (1) patient or parent, (2) provider, or (3) imaging obtained for non-thyroid indications (incidental). Characteristics that were evaluated included rate of malignancy, size of the nodule, location of the nodule, and size of the cancer (if present).

Results

A total of 78 patients with concerning thyroid nodules were analyzed. Within the study, the cancer rate was 27% (21/78), which is comparable to the reported malignancy rate of pediatric thyroid nodules in the literature (22-26%) (4), suggesting that our sample population may be representative of the general pediatric thyroid nodule population. In our study, though the absolute numbers were small, there was a higher rate of malignancy in the incidental group (3/5, 60%) compared to the patient/parent (9/34, 26%) and provider (9/39, 23%) groups. The average size of the thyroid nodule was similar in all 3 groups. The strength of this study was the inclusion of only patients with definitive diagnosis of the thyroid nodule and the possibility of the findings being applicable to the general pediatric population.

Conclusions

In our sample study, incidentally discovered pediatric thyroid nodules had a higher rate of malignancy as compared to those discovered by patients/parents or providers.

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Bone and Mineral Metabolism

BONE AND MINERAL CASE REPORTS I

First Case in Saudi Arabia Revealing Fahr Syndrome Secondary to Hypoparathyroidism: A Case Report

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SAT-357

First Case in Saudi Arabia Revealing Fahr Syndrome Secondary to Hypoparathyroidism: A Case Report

Abstract

Introduction

Fahr Syndrome is a rare inherited or sporadic neurological disorder. It is characterized as an abnormal calcium deposition or in other words, calcification in the brain which almost always occurs bilaterally. Patients with Fahr...