in the ClinVar database (RCV000714711.1) exists from a research lab and was classified as likely pathogenic. Analysis of parental samples showed that the mother was heterozygous for the same genetic variant. She did not have a history of hypoglycemia. Patient was started on diazoxide (8 mg/kg/day) and chlorothiazide with resolution of hypoglycemia. At a follow up visit at 5 months of age, there was no history of hypoglycemia, and no need for adjustments of the diazoxide dose by weight (dose at that time of 7.4 mg/kg/day). **Conclusion:** The ABCC8 reported here is a dominant mutation causing hyperinsulinemic hypoglycemia responsive to diazoxide with a milder phenotype later in infancy. Longitudinal follow up of the case is warranted to understand the long term progress in patients with this particular mutation. **Reference:** Adam MP, Ardinger HH, Pagon RA, Wallace SE, et al. None. 1993. Familial hyperinsulinism.

Adipose Tissue, Appetite, and Obesity

OBESITY TREATMENT: GUT HORMONES, DRUG THERAPY, BARIATRIC SURGERY AND DIET

Non-Alcoholic Fatty Liver Disease Determined by MRI and Its Association with Metabolic Variables in Non-Diabetic Subjects

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MON-597

Non-alcoholic fatty liver disease determined by MRI and its association with metabolic variables in non-diabetic subjects

Background: Non-alcoholic fatty liver disease (NAFLD) is defined as the presence of hepatic steatosis (liver fat accumulation higher than 5% of liver weight) in the absence of other causes. Liver biopsy is recognized as a gold standard for diagnosis, but it is limited by the risks of serious complications. Besides that, the increasing prevalence of NASH led to improved imaging methods, such as Magnetic Resonance Imaging (MRI), that enable quantitative assessment of steatosis by quantifying the hepatic fat fraction (HFF), even with steatosis levels as low as 5.56%.

Objective: The aim of this study was to evaluate the metabolic profile of patients without T2DM according to hepatic steatosis measured by MRI.

Methods: This was cross-sectional study conducted in an Endocrinology Unity in Minas Gerais, Brazil. The study complied with the WMA Declaration of Helsinki and was approved by the Ethical Committee on Human Subject Research from the Faculty Patos de Minas. We recruited non-diabetic subjects aged above 20 years with hepatic steatosis detected by liver sonography. Exclusion criteria were alcohol consumption of more than 20 grams/day for female and 30 grams/day for male, ferritin serum levels above 1000 mg/dL, positive serology for hepatitis B or C and intake of medications known to produce hepatic steatosis. Included subjects underwent HFF quantification by MRI, and the degree of liver fatty infiltration was estimated by using chemical shift imaging. The following biochemical variables were assessed: fasting glucose and HbA1c, HOMA-IR, lipids, AST, ALT and GGT. Analysis: we grouped individuals according to the quartile of HFF and compared clinical and biochemical variables between the groups.

Results: A total of 30 subjects (18 male and 12 females) were included. All subjects were overweight (10% overweight and 90% obese); 7 (23.3%) had 3 criteria and 16 (53.3%) had two criteria for MS. The only variable assessed herein that was different between males and females was HDL-c (40.5 vs 50.5 mg/dL, respectively, p=0.0255). ALT serum levels were significantly higher in subjects in the fourth quartile of HFF, when compared to those in the third quartile (76 vs 47 UI/L, respectively, p=0.037). The other clinical of biochemical variables assessed did not differ between the quartiles of HFF.

Conclusion: our preliminary findings indicate that the biochemical variables related to metabolic homeostasis are poor predictors of the degree of liver fat in overweight non-diabetic subjects. Although screening for NAFLD is still a matter of debate, our results suggest that future discussions about this should take into account that excess body weight per se, independently from biochemical abnormalities, should be considered in the recommendations for screening non-diabetic subjects.

Adrenal

ADRENAL CASE REPORTS I

Aggressive Phenotype Pheochromocytoma Associated with NF-1 and BRCA Mutation

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SAT-222

Introduction

Pheochromocytomas are catecholamine-secreting tumors of the adrenal medulla that occur sporadically or with familial association. Familial predilection occurs in those with multiple endocrine neoplasia type 2, von Hippel-Lindau disease, and neurofibromatosis Type 1 (NF-1). To date, more than 21 gene mutations have been identified that are involved in the development of pheochromocytomas. However, co-existence of two different mutations such as NF-1 and BRCA gene mutations with pheochromocytoma has not been well described.

Clinical Case

A 40 year-old woman with NF-1 and recently diagnosed left breast invasive ductal carcinoma, estrogen positive, progesterone positive, HER-2 negative BRCA2 positive breast cancer and right breast lobular carcinoma in situ underwent staging CT scan. A 2.7 cm left adrenal incidentaloma with heterogeneous texture and 69–100 HU was discovered. Symptom review revealed chronic headaches and increasing frequency of anxiousness and irritability. She had no personal or family history of hypertension. Laboratory analysis showed elevated catecholamine levels included

plasma normetanephrine of 2.8 nmol/L (normal <0.9), metanephrine of 0.9 nmol/L (normal <0.5), 24 hour urine metanephrine 498 mcg/24 hr (normal <180) and 24 hour urine normetanephrine of 1,152 mcg/24 hr (normal <451). Findings were consistent with adrenal pheochromocytoma. She was immediately started on phenoxybenzamine and encouraged to increase fluid intake to expand intravascular volume before surgery. She underwent laparoscopic left partial adrenalectomy fourteen days later. Surgical pathology revealed a 3.0 cm pheochromocytoma in a background of nodular hyperplasia of the medulla, described as an aggressive tumor phenotype based on morphological features.

Conclusions

It has been estimated that 1.5–14% of adrenal incidentalomas are pheochromocytomas. While only 3% of NF-1 patients have pheochromocytomas, 20–50% of NF-1 patients with concurrent hypertension have underlying chromaffin tumors. This association is significant when compared to the pheochromocytoma incidence of 0.1% in all hypertensive individuals. Interestingly, germline mutations of BRCA-1 associated protein-1 gene have been reported in patients with paragangliomas, therefore it may be plausible to consider interactions between NF-1 and breast cancer BRCA-2 gene mutations, resulting in an unusual and more aggressive pheochromocytoma phenotype, even when detected at an early stage, as in this case. It is also important to recognize and adequately treat pheochromocytoma prior to having surgery to avoid possible intraoperative hypertensive crisis, a concept that was underscored in this particular case when the patient was relatively asymptomatic and entirely normotensive preoperatively.

Bone and Mineral Metabolism

PARATHYROID HORMONE TRANSLATIONAL AND CLINICAL ASPECTS

Testing for Parathyroid Hormone: Performances of a Novel Fully Chemiluminescent Automated Assay.

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SAT-387

Background: Parathyroid hormone (PTH) is one of the key regulators of the bone and mineral metabolism. Testing for PTH is essential for the management of hyper- or hypo calcemia. Measurement of PTH is also an important tool for the monitoring of patient with chronic kidney diseases. Our study objective was to determine the performances of a novel fully automated chemiluminescent assay for testing of intact PTH. **Methods:** We evaluated the Maglumi® (Snibe) PTH assay a fully automated two-sites immunoluminometric method based on a solid-phase, paramagnetic microbeads, coated with a monoclonal antibody targeting PTH epitopes. Assay imprecision was assessed with two levels of control materials. Reference values were determined with samples of 24 healthy volunteers. Method comparison was performed with an electrochemiluminescent immunoassay (ECLIA) (Roche diagnostics) with 24 patients' samples. **Results:** The between-run coefficients of variation of the Maglumi® PTH assay were 5.1 and 3.7 % for concentrations of 33 and 992 pg/mL, respectively. The median PTH levels were 121 pg/mL (range: 18 - 369) with the Maglumi assay and 117 pg/mL with the ECLIA method (range: 10 - 482). The upper limit of the reference interval in the healthy volunteers was 63 pg/mL. The correlation between the both methods was good (r=0.87, p<0,001). Passing-Bablok regression analysis showed a slope of 0.96 (95% confidence interval (CI): 0.83 to 1.35) and an intercept of -2.14 (95% CI: -20.49 to -10.47). Bland-Altman plot evidenced a bias between the methods with a mean bias of 4.6 pg/mL. **Conclusions:** Our preliminary data showed excellent concordance and analytical performances for the PTH fully automated immunoassay associated to perspectives of automation and reduction of turn-around time of analysis.

Pediatric Endocrinology

PEDIATRIC SEXUAL DIFFERENTIATION, PUBERTY, AND BONE BIOLOGY

Gonadotropins Levels Measurement in First Morning Voided Urine as a Diagnostic Tool for Central Precocious Puberty

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SUN-097

Background: GnRH stimulation test is the gold standard for the diagnosis of central precocious puberty (CPP). However, it is invasive and costly. Previous studies showed that increased urinary gonadotropins (Gn) level in first morning voided (FMV) urine reflected the integration of elevated nocturnal Gn secretions. Therefore, it could be used to diagnose CPP. Nevertheless, its cutoff value for diagnosis of CPP is limited. **Objective:** To determine the association of Gn levels in FMV urine and serum during pubertal development and establish cutoff value of FMV urinary Gn as an alternative noninvasive method for diagnosis of CPP in girls**. Methods:** Sixty-one girls who had breast development before 8 years of age with sign of rapid pubertal progression (advanced bone age and/or increased height velocity) underwent subcutaneous GnRH agonist test. FMV urinary Gn were also collected on the same day. Both serum and urinary Gn levels were measured using electrochemiluminescence immunoassay (ECLIA) technique. The definite diagnosis of CPP is based on stimulated serum LH > 5 IU/L. FMV urinary Gn were compared between CPP and premature thelarche (PT) groups. The correlation between serum and urinary Gn were assessed and the cutoff value of urinary Gn to diagnose CPP was established. FMV urinary Gn of 480 Thai school girls (control) were also collected to determine the reference values according to their breast Tanner (BT) stages. **Results**: FMV ULH level in girls with CPP was significantly higher than that of PT (2.46 VS 0.8 IU/L; median, P <0.001). However, the level of ULH in PT group was not different from control group with BT1. FMV ULH and ULH: UFSH were well correlated with basal serum LH (r=0.63 and 0.73, respectively, Ps<0.001) and peak serum LH (r=0.44 and 0.54, respectively, Ps<0.001). Base on receiver operating