Pediatric Endocrinology

PEDIATRIC PUBERTY, TRANSGENDER HEALTH, AND GENERAL ENDOCRINE

Diversity of Endocrine Function in Patients with CHARGE Association

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Context: CHARGE association consists of congenital malformation of Coloboma, Heart defect, Atesia choanae, Retarded growth and development, Genital hypoplasia, Ear anomalies and/or deafness. It is often caused by CHD7 gene mutation, which also one of the causative gene for Kallmann syndrome. The endocrine dysfunction in CHARGE association has been reported but not fully understood. Objective: To clarify the mode of growth and frequency of endocrine dysfunction in CHARGE association. Subjective: We investigated the characteristics of growth and puberty, and endocrine function in 23 children (15 males and 8 females, 0~20 years old) with CHARGE association. Results: The birthweight was from -2.74 to +1.14 SDS and the birth length was from -2.86 to +1.10 SDS. 5 children were born small for gestational age. The height below -2SDS in 18 children. GH secretion was evaluated in 11 children with short stature (-9 to -2.3SD) except for one with normal height (-0.3 SD in 6 years old girl); 5 children including one with normal stature were revealed to have GH deficiency. One short girl with GH deficiency previously showed normal GH response to provocation test at 1 year old but has developed to be GH deficient at 7 years old. Gonadotropin-releasing hormone loading tests were performed in 7 males and 3 females. Nine out of 10 children showed hypergonadotropic hypogonadism; one girl showed hypergonadotropic hypogonadism, whose ovaries were undetectable on ultrasound. Human chorionic gonadotrophin (HCG) tests were performed in 6 males with micropenis and/or cryptorchidism. Peak testosterone levels after HCG stimulation were from 0 to 6.99 ng/ml. 4 patients showed peak testosterone levels less than 1 ng/ml. Four boys showed combined gonadotropin deficiency and primary hypogonadism. Conclusions: Our data showed the diversity of endocrine function in children with CHARGE association. GH deficiency can be developed over time. Hypogonadotropic hypogonadism is common, while isolated/combined primary hypogonadism should be taken into consideration in children with CHARGE association.

Bone and Mineral Metabolism

BONE AND MINERAL CASE REPORTS I

Case Series of Ectopic Parathyroid Gland

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SAT-377

The prevalence of mediastinal parathyroid adenoma is unknown. Embryological origin and more extensive aberrant migration of the parathyroid glands result in ectopic glands found in the mediastinum. We report herein 4 cases of ectopic parathyroid adenoma causing primary hyperparathyroidism from three public hospitals in Malaysia.

Case 1: A 70 year old lady with underlying diabetes mellitus, hypertension, chronic immune thrombocytopenic purpura and liver cirrhosis presented with incidental asymptomatic hypercalcemia during an admission for pneumonia. Her blood results revealed high corrected calcium of 2.93 mmol/L (2.02-2.60) and a low phosphate of 0.66 (0.81-1.45) mmol/L with an unsuppressed intact parathyroid hormone (iPTH) of 14.56 pmol/L (1.6-6.9). She had an equivocal urinary calcium excretion ratio of 0.01. Her bone mineral density confirmed severe osteoporosis at distal radius and neck of femur with a Tscore of -3.6 and -3.1 respectively. A hyperfunctioning ectopic parathyroid gland was seen in the Technetium Sestamibi scan which correlates with a mediastinal lymphadenopathy on CECT. The largest node measured 1.6 x 1.2 cm. Parathyroid gland was confirmed on HPE of the video-assisted-thoracoscopic surgical (VATS) excision of the mediastinal mass. Intraoperative iPTH
Avoiding the Heartache: A Case of Familial Hypercholesterolemia

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SAT-566

More than 70% of individuals with atherosclerotic cardiovascular disease are believed to have underlying gene-linked mechanisms leading to hyperlipidemia. It is estimated that 1 in 200 individuals in the United States has heterozygous Familial Hypercholesterolemia (FH). We present a case that highlights the importance of comprehensive care for a patient with heterozygous FH, from screening and risk stratification, to therapy. Our patient is a 43-year-old gentleman with history of hyperlipidemia. At age 25, he was diagnosed with hyperlipidemia and was started on statin therapy. He has strong family history of cardiovascular disease. His mother had her first myocardial infarction (MI) at age 40 and required coronary artery bypass. She also suffered from three strokes. Her maternal aunt and uncle suffered from MIs at age 38 and 40, respectively. Additionally, his maternal grandfather passed away from MI at age 38. The patient’s daughter was found to have total cholesterol level > 300 mg/dL at age 8. He does not have history of obesity, diabetes, previous cardiovascular events, or hypothyroidism. He is athletic and follows a healthy diet. He did not have any xanthomas, xanthelasmas, nor arcus cornealis. At time of initial evaluation, the patient had low-density lip (LDL) level of 180 mg/dL despite therapy with rosuvastatin, ezetimibe, and niacin. Based on these findings, we proceeded with genetic testing. Results of testing showed a heterozygous c.6delG (p.Trp4Glyfs*202) pathogenic mutation of the LDL receptor. We also obtained cardiovascular risk stratification studies. On cardiac CT angiogram, he was found to have extensive, four-vessel disease with 80-90% stenosis of the left ascending artery (LAD) with coronary calcium score of 136 and total score of 219 (99th percentile). Exercise, stress myocardial perfusion scan showed small reversible anteroseptal perfusion abnormality suggestive of mild to moderate ischemia. LAD stenosis was confirmed on a left heart catheter, but no intervention was required. We proceeded with aggressive lipid-lowering therapy with rosuvastatin 40mg daily and alirocumab 300mg monthly. He was also started on aspirin and beta-blocker given coronary artery disease. Following initiation of therapy, the patient’s LDL level dropped to 51 mg/dL with total cholesterol level of 153 mg/dL, HDL of 47 mg/dL, and triglycerides of 109 mg/dL. The patient was encouraged to seek genetic counseling for his children and first degree relatives. His daughter was started on rosuvastatin 7.5mg daily by her pediatrician. The patient has not suffered any cardiovascular events and continues to follow up for therapy. Without aggressive lipid-lowering therapy, the lifespan of FH patients can be significantly shortened. Therefore, identifying FH patients is imperative to prevent cardiovascular disease in these patients and their afflicted family members.

Diabetes Mellitus and Glucose Metabolism

CLINICAL AND TRANSLATIONAL STUDIES IN DIABETES

Endogenous Insulin and C-Peptide Suppression Test Using a Rapid-Acting Insulin Analog in the Diagnosis of Insulinoma

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C-peptide suppression test (CPS) was shown to diagnose the cause of hyperinsulinemic hypoglycemia, i.e. insulinoma, as effectively as supervised 72-hour fast test with less time consuming and cost. In the conventional CPS, regular insulin (RI) is used to induce hypoglycemia that subsequently suppresses endogenous insulin secretion. As RI is measurable in plasma insulin (PI) assay, plasma C-peptide (PCP) but not PI response is therefore used for assessment of endogenous insulin secretion in CPS using RI. As rapid acting insulin analogs (RA) are not measurable in a selected PI assay, both PCP and PI levels can be used to assess endogenous insulin secretion if an RA is used instead of RI in CPS. There is no study on PI and PCP responses to RA in insulinoma. This study aimed to examine efficacy of modified CPS, so-called insulin and C-peptide suppression test (ICPS) by using an RA (insulin aspart) in the diagnosis of insulinoma. Ten patients, 7 with histopathological proven