show significant rises in gonadotropins. Bone age was advanced by more than 1 year. The
patient was started on subcutaneous octreotide with a decrease in IGF-1 to 258 ng/mL after 1
month of therapy. On treatment, linear growth velocity slowed with no interval height gain
over the initial 1-month period; however, the patient's weight continued to increase with a gain
of 1.8 kg. Parents additionally reported hyperphagia, which prompted concern for
hypothalamic obesity in the setting of her known hypothalamic mass. Thyroid function
remained normal on somatostatin therapy. To date, there has been no concern for diabetes
insipidus.
Conclusion: Growth hormone excess may rarely complicate a diagnosis of NF-1 in the setting of
intracranial gliomas. Increased height velocity and/or tall stature for family should raise clinical
suspicion and prompt evaluation. Hyperphagia and significant increases in weight in the setting
of hypothalamic gliomas in patients with NF-1 should raise suspicion for hypothalamic obesity
and prompt lifestyle modifications to curb ongoing weight gain.

Bone and Mineral Metabolism

NEW INSIGHTS INTO PTH AND CALCIUM RECEPTOR SIGNALING

Identification of the First Case of Acquired Autoimmune Parathyroid Hormone (PTH) Resistance Due to PTH1 Receptor (PTH1R) Autoantibodies

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Background: Here we describe a patient who presented with symptomatic hypocalcemia and a biochemical picture suggestive of PTH resistance. PTH resistance is a hallmark of pseudohypoparathyroidism, a heterogeneous group of rare disorders caused by genetic or epigenetic alterations of PTH/PTHrP signaling. However, PTH receptor-related autoimmune etiology has not been identified as the underlying mechanism for PTH resistance. Here we describe the first case of acquired autoimmune PTH resistance that is secondary to PTH1R autoantibodies.
Clinical Case: A 60-year-old African-American woman, who previously had normal calcium homeostasis, presented with acute, symptomatic hypocalcemia, hyperphosphatemia and markedly elevated serum PTH, consistent with parathyroid hormone resistance. She did not have other hormone resistance or a clinical phenotype suggestive of pseudohypoparathyroidism. Whole-exome sequencing and *GNAS* methylation analysis revealed no genetic or epigenetic defects of the PTH/PTHrP signaling pathway. Treatment with Calcitriol and Calcium supplements was initiated with good clinical response. Within 10 years of follow-up, the patient developed autoimmune hypothyroidism, alopecia and an unusual form of membranous glomerulonephritis, raising the suspicion for an autoimmune etiology for PTH resistance. Luciferase immunoprecipitation system assay identified antibodies against PTH1R with mapping to the N-terminal extracellular ligand-binding domain (amino acids 1- 178). Using an in vitro biological assay in GP-2.3 cells, we found that the antibodies derived from the patient's serum blocked PTH downstream signaling via $G_s$alpha/cAMP/protein kinase A pathway in a concentration-dependent manner.
The patient's autoantibody profile led to the diagnosis of additional autoimmune diseases, including atrophic gastritis and Sjogren syndrome. Lymphocyte immunophenotyping using flow cytometry revealed an overall normal B and T cell profile, but with decreased frequencies and numbers of switched and non-switched memory B cell subsets and an increased frequency and number of the $CD8^+$ naïve cell population. Genes associated with autoimmune inflammatory disorders were sequenced but no pathologic changes were detected.
Conclusions: Identification of the first case of autoimmune PTH resistance secondary to PTH1R autoantibodies extends the etiologic spectrum of hypoparathyroidism and should be considered when a patient presents with findings consistent with pseudohypoparathyroidism, especially in the presence of additional autoimmune diseases.

Pediatric Endocrinology

PEDIATRIC ENDOCRINE CASE REPORTS I

A Novel De Novo GATA3 Gene Mutation in an Adolescent with HDR Syndrome

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Background: *GATA3* encodes a transcription factor critical for embryonic development of the parathyroid glands, kidney, inner ear, thymus, and the central nervous system. Heterozygous loss-of-function mutations in *GATA3* are associated with hypoparathyroidism, sensorineural deafness and renal disease (HDR syndrome). **Clinical Case:** A 12 yo male with left hip pain underwent a closed reduction for left slipped capital femoral epiphysis. The pre-op evaluation revealed hypocalcemia (serum Ca 7.7 mg/dL; nl: 8.8-10.2), creatinine 0.46 mg/dL (0.5-1.0), TSH 3.16 uU/mL (0.3-4.2), FT4 1.36 ng/dL (0.8-1.8). Oral calcium and vitamin D supplementation was begun, and 2 wks later, follow-up evaluation revealed serum Ca of 9.4 mg/dL, intact PTH 4.6 pg/mL (10-69), phosphorus 5.9 mg/dL (3.3-5.3), 25-OHD 26 ng/mL (30-100), and a normal chromosomal microarray. Bone density (DXA) Z-scores for hip and spine were -1.7 and 0.8, respectively. At age 13 he underwent bilateral osteotomy due to bilateral hip dysplasia and removal of hardware the next year. At age 15 he underwent left total hip replacement for avascular necrosis. In the post-operative period hypocalcemia recurred (5.9-6.7mg/dL), and he was referred for endocrine evaluation. He was of mixed African American