Bone marrow aspiration was normal. We could not perform flow cytometry of anti-neutrophil cytoplasmic antibodies (C-ANCA). Hyperthyroidism persisted and a second I-131 treatment was performed (20 mCi) in June 2018. A month later she presented hypothyroidism, levothyroxine was indicated. She continued with episodes of febrile neutropenia until March 2019, 23 months after the diagnosis of hyperthyroidism, 16 months after stopping methimazole and 8 months after having initiated levothyroxine treatment and having normal thyroid levels.
Conclusion: We presented a young female patient with persistent and recurrent neutropenia despite having stopped methimazole, and regardless of her thyroid hormone levels. Although neutropenia usually appears in the first months of treatment, it seldom occurs much later and almost never after stopping the drug. We could not reach an etiological diagnosis of neutropenia, but it is probable that methimazole had triggered an immune-hematological illness associated to Graves' disease.

Tumor Biology

ENDOCRINE NEOPLASIA CASE REPORTS II

Susceptibility Genetic Testing and Functional Imaging Modalities in the Management of Bladder Paragangliomas

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Introduction: Bladder Paragangliomas (PGLs) are rare neuroendocrine tumors derived from sympathetic paraganglionic tissue within the bladder wall, accounting for <1% of all Pheochromocytomas and Paragangliomas (PPGLs). >40% of PPGLs are associated with inherited syndromes through mutations affecting citric acid cycle enzymes (commonly SDH). Susceptibility gene identification has important implications for long-term care and facilitates targeted cascade genetic screening. Functional imaging using MIBG, Gallium DOTATATE and FDG-PET have become important tools in both diagnosis and treatment (Peptide Receptor Radionuclide Therapy).
Clinical Cases: We report the demographics, clinical characteristics and novel features of 7 patients with bladder PGLs. The series includes 2 females and 5 males, median age 38 years (range 14-68). 5 presented with hematuria and 2 were detected incidentally (1 found on radiological imaging and the other during cystoscopy surveillance). Other symptoms reported were headaches, sweating and palpitations which were relieved by urination. Only 1/7 had a known family history of PGLs. 5/7 patients had elevated plasma normetadrenaline levels and 2 had non-elevated catecholamine metabolites (these 2 patients were asymptomatic).
6/7 patients had genetic testing performed and pathogenic variants were identified in 4 (Fumarate hydratase (FH), SDHA, SDHB*2 genes) and no pathogenic variant identified in 2 patients in our genetic panel of 10 PPGL genes. All primary tumors demonstrated MIBG avidity and in 2 patients assessed there was PGL FDG-PET avidity. Metastatic disease was present in 2 patients (2 SDHB mutations; with 1 MIBG avid bone and 1 FDG-PET avid nodal metastasis). SDHB immunostaining on resected histology was available for 3 cases - absent SDHB immunostaining in the patient with SDHA mutation and strongly positivity in 2 patients (1 with no genetic mutation and in 1 with FH mutation).
Conclusions: The majority (>65%) of patients with bladder PGL have a germ line mutation in a susceptibility gene involving the citric acid cycle. An extended gene panel should be performed in all patients diagnosed with bladder PGLs including SDHA and FH gene mutations. SDH immunostaining of tumour can indicate SDHx gene defects but can be normal in FH mutations. SDHB is associated with increased risk of malignant/metastatic behavior. All 3 modalities of functional imaging (Ga DOTATATE, FDG PET, & MIBG) have a role in the assessment and treatment decision making in the management of bladder PGLs.

Bone and Mineral Metabolism

BONE DISEASE FROM BENCH TO BEDSIDE

Low Dose Ethinyl Estradiol in Women with Cystic Fibrosis Does Not Preserve Bone Mass

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BACKGROUND: Cystic fibrosis-related bone disease (CFBD) affects 26% of adults with cystic fibrosis (CF). CFBD increases the risk for fractures, which in turn limits patients' ability to effectively perform daily therapies necessary to maintain health. Factors contributing to CFBD include nutritional deficiencies, inflammation, glucocorticoid use, CF-related diabetes and untreated hypogonadism. Hypogonadism in CF is thought to be functional, although the distribution of etiologies of female hypogonadism in the modern era of CF therapies is unknown. Estrogen supplementation is commonly prescribed in the form of oral contraceptives to women with low bone mineral density (BMD). At our CF center, the average dose of ethinyl estradiol prescribed to women is 20 mcg. Recent evidence suggests that oral estrogen is ineffective for restoring bone health in women with functional hypogonadism and specifically that doses < 30 mcg oral ethinyl estradiol are inadequate. It is unknown if estradiol supplementation will restore and/or maintain BMD in women with CFBD.
METHODS: The purpose of this study was to examine the skeletal health of a cross-section of premenopausal women seen at a single CF center taking 20 mcg or less of ethinyl estradiol daily (low-dose estrogen) compared to women not taking estrogen supplement. As screening for an IRB-approved intervention study, we collected health information by chart review.