Discussion: We present a case of elevated FGF23 with renal phosphate wasting concerning for TIO. This case represents the diagnostic dilemma of elevated FGF23 as well as difficulty in discerning the source in TIO. Although exact tumor source remained unclear in our patient, adipose tissue in the thoracic inlet was a potential culprit. Elevated FGF23 leads to urinary phosphate wasting and inhibition of 1α-hydroxylase. Daily urine phosphate excretion > 100 mg or FEPO4 > 5 % strongly suggest renal phosphate wasting. TIO and iron-induced hypophosphatemia are two most common acquired causes of high FGF23. TIO is mostly caused by benign soft tissue and bone tumors. It can take years to establish the diagnosis due to small size and obscure location of the tumors. Advanced investigation with FDG-PET, ^{68}Ga-DOTATATE scan, octreotide scan and venous sampling of FGF23 can aid in diagnosis. Tumor resection is usually curative. Burosumab (anti-FGF23 monoclonal antibody) can be considered when the tumor is not amenable to resect.

Bone and Mineral Metabolism

BONE AND MINERAL CASE REPORT

Early Osteoporosis in RYR1-Related Central Core Disease

Ezza Tariq, MBBS[1], Lubna Mirza, MBBS[2].
[1]Nishtar Medical College, Multan, Pakistan, [2]Norman Endocrinology Associates, Norman, OK, USA.

Background: Central core disease(CDC) is a congenital neuromuscular myopathy with a wide range of phenotypic presentations, ranging from delayed motor development, frequent falls, and difficulty maintaining posture. CDC is a rare presentation of RYR1 (Calcium release channel gene) mutation, which is also linked with the etiology of malignant hyperthermia. **Clinical Case:** We present a case of a 57-year-old woman who was diagnosed with osteoporosis at age of 52 with a T score of -2.3 after she had a fragility fracture of the knee. She suffered from multiple falls from poor balance. Her most recent DXA bone density scan from December of 2018 showed a T score of -2.6. On genetic testing, she was found to have a RYR1 heterozygous mutation, on exon 28.c.3800C to G(p.P1267 Arg). This sequence change led to the replacement of proline with arginine at codon 1267 of RyR1 protein. None of her immediate and extended family members showed any signs of CDC. We assume that the loss of sufficient muscle strain on bone and the catabolic effect of RYR1 myopathy are major causes of osteoporosis in our patient, although menopause, personal history of smoking, and alcohol intake could also be contributing factors. Teriparatide along with daily Calcium and Vitamin-D was prescribed. Later on, denosumab injection was also added to the regimen. The patient still has at least one episode of unprovoked fall in a month, but luckily she has not had any more fractures. **Conclusion:** To our knowledge, this is the first case where early osteoporosis in RYR1 myopathy has been reported.

Bone and Mineral Metabolism

BONE AND MINERAL CASE REPORT

Ectopic Atypical Parathyroid Adenoma Presenting With Pancreatitis

Paul Shiu, DO[1], Caroline Tashdjian, MD[1], Tarandeep Kaur, MD[2].
[1]St. joseph medical center, Stockton, CA, USA, [2]Dignity Health, St joseph Medical Center, Stockton, CA, USA.

Background: Atypical Parathyroid Adenoma (APA) and parathyroid carcinomas (PC) are rare parathyroid tumors (<1%) causing primary hyperparathyroidism. We present a case of ectopic APA in the left anterior mediastinum. **Case:** A 38 year old male with hyperlipidemia admitted for acute pancreatitis noted to have calcium level of 13.4 mg/dl (8.6–10.3mg/dl) in the absence of family history of hypercalcemia or kidney stones. He was treated with zoledronic acid, intravenous fluids and calcitonin for hypercalcemia. Pancreatitis resolved with fluid management. Calcium on discharge was 9.9 mg/dl. 8 week follow-up labs showed PTH of 420 pg/ml (14–64 pg/ml), calcium 10.2 mg/dl, phosphorus 1.9 mg/dl (2.5–4.5 mg/dl), vitamin D 19 ng/ml (30–100 ng/ml), 24-hour urinary calcium 115 mg/24 hour (55–300 mg/24 hour) and calcium to creatinine ratio 0.41. Hence diagnosis of primary hyperparathyroidism was made but given the degree of calcium and PTH elevation there was a concern for PC. Ultrasound of the neck demonstrated no abnormality. Sestamibi scan showed irregular focal accumulation of radiotracer in the left upper mediastinum suggestive of mediastinal ectopic parathyroid adenoma (EPA). CT chest with contrast showed a 3.9 x 2.2 x 3.4cm lobulated, heterogenous, hypodense, minimally enhancing mass in the left anterior prevascular space. Diagnosis of EPA was made and he underwent left video assisted thoracic surgery. Operative findings showed a multilobular mass at the level of the aortic arch between vagus and phrenic nerves. Intraoperative PTH went from 1124 pg/ml pre-incision to 160 at 15-minute post-excision. Postoperative calcium was 9.6 mg/dl and PTH 51 pg/ml. Final pathology showed 3.7 x 2.5 x 2cm, hypercellular parathyroid with prominent fibrous band and parafibromin retention compatible with APA. **Discussion:** APA is an intermediate form of parathyroid neoplasm with uncertain malignant potential, showing atypical histological features without evidence of invasive growth. This poses a diagnostic challenge of PC as the histopathological features overlap and requires an expert pathologist to make the diagnosis. Inactivating CDC73 mutations, encoding parafibromin, is the most common genetic abnormality. Loss of parafibromin has been seen in 100% cases of PC and predicts poor prognosis. Due to rarity of the disease there are no clear guidelines on follow up. Based on literature, annual follow up is suggested with biochemical testing and imaging for the first 5 years after surgery and every 2–3 years thereafter due to 3% recurrence rate and potential progression to PC. Cetani et al in 2019 reported 672 cases of APA, only 3 were ectopic in the mediastinum thus making our case rare. APA is a rare entity as biochemical profile and histopathological features overlap with PC and can be misdiagnosed. Greater awareness of APA may improve capture of cases, allowing the development of guidelines to recognize and treat it.

Bone and Mineral Metabolism

BONE AND MINERAL CASE REPORT

Ectopic Intact PTH Secretion Causing Humoral Hypercalcemia of Malignancy