was delayed due to poor functional status and concurrent discovery of an EBV-positive nasopharyngeal carcinoma. Prior to surgery patient was treated with phosphorus and calcitriol supplements. Post-operatively serum phosphorus and FGF-23 levels were normalized. Patient also improved clinically. Patients treatment course was complicated by secondary hyperparathyroidism; however, this improved following surgery. Conclusion: Diagnosis of TIO can be delayed due to its nonspecific symptoms. Thus, in patients with chronic bone pain, muscle weakness, and atraumatic fractures, TIO should be kept on the differential and these patients should undergo thorough biochemical and imaging evaluation. Tumor localization could be challenging. Patients should be managed with supplements of active vitamin D and phosphorus with goal to normalize phosphorus level to prevent further bone demineralization prior to surgery. However, surgical intervention remains the mainstay of management as this is curative of TIO.

Bone and Mineral Metabolism
BONE AND MINERAL CASE REPORT

Successful Early Treatment of Severe Neonatal Hyperparathyroidism With Cinacalcet

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Introduction: Neonatal severe hyperparathyroidism (NSHPT) can cause significant bone disease early in life and prompt treatment is therefore necessary. Cinacalcet is a calcimimetic primarily used in adult patients with hypercalcemia to treat secondary hyperparathyroidism, but has only been trialed in neonates with NSHPT. Successful treatment appears to be dependent on the mutation leading to hypercalcemia. We describe a neonate born with severe hypercalcemia and a suspected mutation of the calcium sensing receptor (CasR) who received successful early treatment with cinacalcet and decrease in calcium levels.

Case Report: A full-term baby girl was found to have rapid breathing at 2.5 hours of life requiring CPAP. A chest x-ray demonstrated bone demineralization with rib fractures. Lab evaluation demonstrated hypercalcemia (total calcium = 12 mg/dL) with an inappropriately elevated PTH level of 386 pg/mL, hypophosphatemia (3.3 mg/dL), normal magnesium (2.0 mg/dL), a normal urine calcium to creatinine ratio of 0.26 and calcium to creatinine clearance ratio of 0.05 and a slightly low vitamin D-25 of 28.8 ng/mL.

The father had a history of asymptomatic hypercalcemia without a diagnosis. Paternal genetic testing identified a heterozygous pathogenic CASR defect: c. 554G>A (p.Arg185Gln). This has been described in patients with NSHPT.

The patient was initially treated with IV fluids and Lasix, but calcium levels did not decrease. Cinacalcet therapy was given on day of life 10. Patient had a decreased PTH to 231 pg/mL after one day. After 26 days of treatment, patient’s PTH level decreased to 63 pg/mL. Patient was weaned off of CPAP and was discharged home.

Discussion: Cinacalcet, a calcimimetic that works at the level of the CASR, was able to successfully and significantly decrease PTH levels in a neonate patient with NSHPT. Treatment options are limited in patients with this condition and we believe prompt treatment with this therapy facilitated patient’s discharge. The patient’s osteopenia secondary to the NSHPT and subsequent rib fractures resulted in a prolonged requirement of CPAP. Early recognition and treatment, even prior to results of genetic testing, prevented further fractures. We demonstrate the potential benefit of calcimimetics in a case of NSHPT where conventional treatment was ineffective. More importantly, we anticipate improvement in osteopenia and any future comorbidities secondary to this condition. Continued success with this treatment is yet to be evaluated.

Bone and Mineral Metabolism
BONE AND MINERAL CASE REPORT

Successful Medical Management of a Non-Localising Case of Tumour-Induced Osteomalacia

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Introduction: TIO is a rare paraneoplastic syndrome characterised by renal phosphate wasting due to fibroblast growth factor-23 (FGF23) over-secretion from a phosphaturic mesenchymal tumour (PMT). While surgery is potentially curative, localisation is often challenging.

Clinical Case: A 53 year old lady presented with recurrent fragility fractures in the spine (T10-L1, L4, S1-2), right femoral neck fracture and pelvic fractures at the bilateral superior and inferior pubic rami, associated with a 2 year onset of lower limb pain and proximal myopathy. Power was 2/5 proximally, rendering her progressively chairbound. She had no family history of fragility fractures. Biochemistry revealed hypophosphatemia of 0.48 mmol/l (NR 0.86–1.45 mmol/l), normal adjusted calcium of 2.32 mmol/l (NR 2.15–2.55 mmol/l), hyperphosphaturia (TmP/GFR 0.39 mmol/ml, NR 0.88–1.42 mmol/ml), mildly insufficient 25(OH)D level of 25 µg/l, inappropriately suppressed 1,25(OH)2D at 13 pg/ml (NR 18–78 pg/ml) and raised FGF23 at 484 RU/ml (NR<180 RU/ml). Localisation of the PMT was unsuccessful, despite multiple investigations including 68-Gallium-DOTANOC PET-CT, bilateral lower limb MRI for non-specific inguinal lymph nodes and various ultrasonographic evaluation of soft tissue lesions in including biopsy of a benign breast tumour. Surgical removal of the breast papilloma did not affect FGF23 levels. In the absence of any suspicious lesion, selective venous sampling was not performed due to uncertain utility. She was treated medically, requiring 16mmol oral phosphate, 1000 IU cholecalciferol and 0.5mg calcitriol daily, with a view to perform interval DOTA-peptide scan. Despite an increase in FGF23 to 760 RU/ml over 29 months, phosphate level was maintained in the low-normal range and alkaline phosphatase, as a marker of disease activity, normalised from 370 U/l to 92 U/l (NR 40-130U/l). Development of secondary hyperparathyroidism improved with uptitration of calcitriol. There was no hypercalcuria on monitoring of