mineralization. There were no neutralizing antibodies. No treatment-emergent adverse events led to study or treatment withdrawal. Serum phosphorus was maintained with long-term burosumab treatment, with no evidence of loss of effect in adults with XLH. Burosumab dose reductions effectively managed mild hyperphosphatemia. Frequency, severity, and types of AEs reported were consistent with previous burosumab trials.

Thyroid

THYROID DISORDERS CASE REPORTS I

Use of Plasmapheresis for Treatment of Thyroid Storm Resistant to Antithyroidals

Myrian Noella Vinan Vega, MD[1], Ana Marcella Rivas Mejia, MD[2], Kenneth Nugent, MD[1].
[1]TEXAS TECH HEALTH SCIENCE CENTER, LUBBOCK, TX, USA, [2]Texas Tech Univ Hlth Sci Ctr, Lubbock, TX, USA.

SUN-515

Management strategies of thyroid storm include measures to reduce thyroid hormone synthesis, hormone release, conversion from T4 to T3, and inhibition of the peripheral effects of excessive thyroid hormone. Plasmapheresis has been described as a treatment option when traditional therapy is not successful or not feasible.

We present a case of an adult patient who presented in thyroid storm in whom plasmapheresis was used successfully as a bridge to thyroidectomy.

51-year-old female with history of hypertension presented with sudden onset shortness of breath, and palpitations. She had an irregular heart rate of 140BPM, respiratory rate of 40, mean arterial blood pressure 65mmHg. Electrocardiogram confirmed atrial fibrillation with rapid ventricular response and the patient was admitted to the intensive care unit. She was started on diltiazem drip and subsequently received amiodarone and electrical cardioversion due to persistent rapid heart rate. She developed respiratory distress and required endotracheal intubation.

Initial thyroid profile revealed low TSH and normal FreeT4, but her FT4 increased above normal on day 2, treatment for thyroid storm was initiated with potassium iodine, hydrocortisone and propylthiouracil. She was kept on propranolol 80 mg q/4h, intravenous esmolol 50 mcg/kg/min, diltiazem drip 5 mg/hr, and was started on Digoxin 0.25 mg q/4h. TSI and TPO were undetectable, and thyroid ultrasound revealed a right nodule measuring 5 x 2.2 x 3.7cm. Thyroid storm was attributed to a toxic nodule exacerbated by exposure to excess iodine (contrast for imaging and amiodarone). Propylthiouracil, hydrocortisone and beta blocker were maximized, and cholestyramine was added, but their heart rate remained elevated, blood pressure worsened requiring synchronized cardioversion.

Because of persistent hyperthyroid state, refractory to medical treatment, patient was started on plasmapheresis on day 10 of hospitalization, she underwent 5 sessions with significant reduction in Free T3 and Free T4 (Figure 1), and remarkable improvement in her hemodynamic status and resolution of tachycardia. Patient underwent total thyroidectomy on day 16, without complications.

Plasmapheresis has been described as a treatment option for refractory thyroid storm, as a bridge therapy prior thyroidectomy. During plasmapheresis, thyroid-binding globulin, thyroid hormones, cytokines and putative antibodies are removed with the plasma; then the colloid replacement provides new binding sites for circulating free thyroid hormone (2). Although albumin binds thyroid hormone less avidly than TBG, it provides a much larger capacity for low-affinity binding that may contribute to lower free thyroid hormone levels, providing a window for thyroidectomy.

1.
Muller C et al, Role of Plasma Exchange in the Thyroid Storm, Therapeutic Apheresis and Dialysis15 (6): 522–531

Bone and Mineral Metabolism

BONE AND MINERAL CASE REPORTS I

Identification of Heterozygous LRP5 Mutation and a TGFβ-1 Variant of Unknown Significance in a Patient with Hearing Loss, High Bone Mass, and Oropharyngeal Exostoses

Cheng Cheng, MD[1], James Avery, MD[1], Gary Gottesman, MD[2], Steven Mumm, PhD[3], Michael P. Whyte, MD[2].
[1]WASHINGTON UNIVERSITY SCHOOL OF MEDICINE, St Louis, MO, USA, [2]Shriners Hospital for Children, Saint Louis, MO, USA, [3]BARNES JEWISH HOSPITAL, Saint Louis, MO, USA.

SAT-384

Background: Manifestations of the high bone mass (HBM) disorders not only include strong bones, but also excessive bones causing cranial nerve palsies and oropharyngeal exostoses. Due to overlap of clinical phenotype in dense bone diseases, one or more distinct genes may be involved. Up-regulation in bone formation can result from gain of function mutation of the low-density lipoprotein receptor-related protein 5 (LRP5), which mediates activation of the canonical Wnt pathway via co-binding with the frizzled protein, but may also be a consequence of activating mutations in the transforming growth factor β1 (TGFβ-1) gene that associate with stimulated osteogenesis. **Clinical Case:** Here we report a 41 year-old woman referred for incidentally found dense bones on screening dual-energy X-ray absorptiometry (DXA) that led to subsequent revelation of several family members sharing similar histories including inability to float in water, strong bones on skeletal surgery, and presence of palatal exostoses. Her childhood history included mandibular pain developing at age 15 years due to bony overgrowth of her lower jaw requiring multiple drilling for removal. At age 33, she manifested trigeminal neuralgia initially responsive to medical management but eventually needed microvascular decompression for unremitting pain. Preoperative brain magnetic resonance imaging (MRI) noted significant hyperostosis of the skull as well as mild narrowing of internal auditory canals, for which auditory testing showing mild mixed hearing loss in her right ear. Skeletal survey revealed diffuse thickening of axial and appendicular skeleton with characteristic endosteal hyperostosis. DXA demonstrated Z scores of +8.3 and +5.3 in the lumbar spine and total hip, respectively. Torus palatinus was also identified on exam. Mutational analysis disclosed a heterozygous LRP5 missense mutation, c.844A>G, p.Met282Val,

together with a variant of unknown significance in TGFβ-1 (c.887G>A, p.Arg296Gln) possibly linked to Camurati-Engelmann disease (progressive diaphyseal dysplasia). As transmission in both conditions follows an autosomal dominant pattern, multigenerational family history was elicited from patient that her mother, who underwent knee replacement in her 80's, was described by the surgeon as "having bones of a 30-year old man," and that her deceased maternal grandmother possibly had "thickened" bones. Half of patient's 18 siblings reportedly also carry features suggestive of high bone mass phenotype. Genetic testing of family members is planned. **Conclusion:** While HBM disorders protect against fractures, it is important to recognize that these relatively benign conditions may present with neurological and oropharyngeal complications prompting the need for their surveillance and surgical precautions during orthopedic procedures.

Thyroid

THYROID DISORDERS CASE REPORTS II

Thyroid Disease And Infrared Imaging Of Eyelids

Shannon Luu, BS[1], Gloria Wu, MD[2], Brian Leung, BS[3], Donia Momen, BS[4], Chap-Kay K. Lau, BS[5].

[1]Santa Clara University, Santa Clara, CA, USA, [2]University of California, San Francisco, San Francisco, CA, USA, [3]Tufts University, Medford, MA, USA, [4]San Jose State University, San Jose, CA, USA, [5]University of California, Irvine, Irvine, CA, USA.

SAT-513

Background: Thyroid eye disease is thought to present as proptosis and/or severe conjunctival chemosis. Severe dry eye disease and its symptoms of non-specific eye pain and foreign body sensation in the eye can be overlooked as an early biomarker of thyroid disease. New infrared imaging can be used to evaluate dry eye and eyelid gland anatomy. Infrared imaging and detailed history of thyroid eye symptoms may lead to subsequent testing of thyroid function and more referrals to thyroid specialists.

Purpose: Using infrared photography to evaluate dry eye complaints in patients: do they have thyroid disease?

Methods: A retrospective chart review (2017–2019) of patients with dry eyes, eyelid imaging with infrared photography and thyroid lab testing was performed. Infrared photography with 820 nm wavelength (Heidelberg Spectralis, Heidelberg, Germany). Percentage loss of Meibomian glands was identified for each eye, then analyzed, per patient. The control population consisted of patients with no dry eye complaints, no thyroid testing or thyroid history. Exclusion criteria: patients over the age of 90 years and patients with a history of glaucoma, diabetes, cataract surgery, and eyelid surgery. Age matching was done (±5 years).

Results: n=48 patients, avg age=57.73 years (sd=16.81, range 21–85 years). Thyroid patients: n=24 patients, male=10, female=14, avg age= 57.12 years (sd=16.65, med=55.5, range 23–83 years). Controls: n=24 patients, male=9, female=15, avg age=58.33 years (sd=17.30, med=58, range 21–85 years). Loss of Meibomian glands: thyroid=40.94%, control=5.10% ($p<0.0001$, t-test). Dry eye complaints: thyroid = 16/24, control = 0/24 ($p<0.0001$, x^2).

Discussion: Meibomian glands are glands in the upper and lower eyelids. These glands provide the lipid component of the tear film, thus slowing the evaporation of the tears and stabilizing the tear film with each blink. Meibomian gland loss would explain the dry eye symptoms in an abnormal thyroid patient population. Infrared photography can be performed with a #87 camera lens filter (cost = $65). The loss of Meibomian glands may be an early sign for thyroid disease.

Conclusion: Infrared photography may be helpful in identifying severe dry eye, thus leading to increased awareness of thyroid eye disease symptoms in our patients in ophthalmology, endocrinology, and primary care.

Adrenal

ADRENAL CASE REPORTS I

Eletriptan (Relpaxa$^{Tm)}$ Causing False Positive Elevations in Urinary Metanephrine

Zachary Bloomer, MD[1], Nath Priti, MD[1], Thanh Duc Hoang, DO[2], Mohamed K.M. Shakir, MD[3].

[1]Walter Reed National Medical Center, North Bethesda, MD, USA, [2]Walter Reed National Medical Center, Bethesda, MD, USA, [3]Walter Reed National Military Medical Center, Bethesda, MD, USA.

SAT-192

Background:

The diagnosis of pheochromocytoma depends crucially on the demonstration of excessive production of catecholamines. This step, however, is fraught with several difficulties, in particular with false-positive test results. Drugs such as phenoxybenzamine and tricyclic antidepressants are the most frequently associated causes for false-positive results. Other medications are also known to cause a false positive elevation of urinary metanephrines. We are reporting a patient with markedly elevated urine metanephrines associated with the use of Eletriptan hydrobromide (Relpaxa^{TM}), a drug commonly used for treating migraine.

Clinical Case:

A 29-year-old man with a history of migraine managed on ibuprofen and recently started Eletriptan presented to the emergency room complaining of a 24-hour history of progressively worsening headaches. At the time of initial evaluation his blood pressure was in the 220s/160s with a creatinine of 1.9 mg/dL with unknown baseline. He was managed on an IV nicardipine drip. Due to his young age he underwent an evaluation for secondary causes of his hypertension. Laboratory: normal aldosterone/renin level (ratio was 0.4), normal midnight salivary cortisol and normal thyroid function studies. Urine screening for drug abuse was also negative. A 24-hour urine metanephrine level, while the patient was taking Eletriptan, was markedly elevated (normetanephrine 1341mcg (ref 82–500) and metanephrine level of 2494 mcg (ref 45–290). In contrast, the plasma metanephrines were only mildly elevated (metanephrines level 27 pg/ml (ref 0–62) and normetanephrine level of 255 pg/ml (ref 0–145)). Adrenal CT did not reveal any evidence of adrenal nodules. Additionally a Gallium-68 PET/CT scan did not reveal any evidence of pheochromocytoma or paraganglioma. Eletriptan was discontinued and his blood pressure was controlled on oral medications. Within one week of stopping Eletriptan his urine metanephrines (metanephrine 76 mcg/ 24 hrs, normetanephrine 277 mcg/dL) and plasma metanephrines (metanephrine 39 pg/mL, normetanephrine 148 pg/mL) normalized.