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Introduction: Primary trimethylaminuria (TMAU), also known as fish malodor syndrome is a rare condition that is characterized by trimethylamine excess. The hallmark of this condition is a body malodor similar to decaying fish. As this metabolic disorder is uncommon, this case highlights the management options for an endocrinologist.
Case presentation: A 56-year-old man was referred to the endocrine clinic for TMAU. Starting in puberty, the patient's family and friends noted a malodor that he has never been able to detect. After several decades, the patient was diagnosed clinically by a dermatologist. Since then, the patient's management had included avoiding cholines in his diet, which included egg yolks and salt-water fish. Additionally, for severe episodes occurring once to twice a year, he took a cup of charcoal daily plus Metronidazole 500 mg twice a day for ten days. The patient was the only member of his family with this condition. Physical exam was unremarkable expect for a faint malodor. Metronidazole, charcoal, and a genetics consult were ordered.
Discussion: TMAU is a rare metabolic disorder in which an individual is not able to convert trimethylamine into trimethylamine N-oxide due to a defect in the hepatic oxidase system. It results from a mutation in the flavin-containing monooxygenase 3 gene (FMO3) that is inherited via autosomal recessive pattern. An excess excretion of trimethylamine in the urine, breath, sweat, and reproductive fluids results in a body malodor similar to that of decaying fish. The odor may be exacerbated by increase in body temperature, emotional changes, puberty, and prior to and during menstruation in women and often results in distressing psychosocial difficulties. Thus, early institution of dietary and pharmacological measures will likely have a major impact on quality of life.
Treatment options are limited and include topical, dietary, and medications. Topical approaches include antiperspirants, deodorants, and pH-balanced soap. A diet low in choline-containing foods such as dairy, beans, and marine fish is beneficial as choline is metabolized to trimethylamine by the intestinal bacteria. For severe cases, antibiotics such as Metronidazole, Rifaximin, and Neomycin sulfate are helpful to reduce the intestinal bacterial load. Other therapeutic strategies consist of activated charcoal (750 mg twice a day for ten days) and copper chlorphyllin (60 mg three times a day for three weeks); both reduce urinary free trimethylamine and increase the concentration of trimethylamine N-oxide.

Cardiovascular Endocrinology

CARDIOVASCULAR ENDOCRINOLOGY AND LIPIDS DISORDERS CASE REPORT

A Case of Hypolipidemia and Hypocholesterolemia; Cause and Consequences

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Background: Hypolipidemia and hypocholesterolemia are uncommon and because of the established risk of hypercholesterolemia for cardiovascular disease, reduced lipids and total cholesterol levels are often clinically desired and/or deemed clinically inconsequential. A finding of persistently low lipid levels and total cholesterol may however not be innocuous nor desirable. We describe the case of a 46 yr old man with persistently low total cholesterol levels <70mg/dl and the associated complications and comorbidities identified. **Clinical Case:** A 46 yr old Hispanic man with non-alcoholic fatty liver disease (NAFLD) was referred for evaluation of hyperhidrosis in the setting of persistent hypolipidemia and hypocholesterolemia. Review of the patient's clinical and biochemical history showed persistently low total cholesterol (mean 58mg/dl), hypotriglyceridemia, and low LDL-C (mean 13.4mg/dl) over the prior 7 yrs in addition to undetectable serum lipoprotein A. Evaluation for secondary causes of hypolipidemia, such as multiple myeloma, was unremarkable. He was found to have low carotene, borderline vitamin A and low vitamin E levels while the rest of his serum fat-soluble vitamins were normal. His mother who had presumed Alzheimer's dementia also had a history of very low cholesterol levels. The degree and persistence of his hypolipidemia and hypocholesterolemia raised the possibility of a genetic etiology of his hypolipidemia. Genetic testing confirmed that the patient was heterozygous for a pathogenic variant in the APOB gene, consistent with familial hypobetalipoproteinemia (FHBL) which is autosomal recessive linked. Subsequent close review of his clinical history revealed other potential complications and comorbidities of FHBL including NAFLD with prediabetes, hypogonadism, progressive cognitive and memory decline, peripheral neuropathy and multiple neuropsychiatric syndromes including adult ADHD, borderline personality disorder, bipolar disorder and chronic anxiety. He is presently on vitamin E and A supplementation and being followed by neurology and psychiatry in addition to ongoing endocrine and metabolic clinical surveillance. In addition, in view of his maternal history and several biologic children he has undergone formal genetic and family counselling. **Conclusions:** While lipid panels are ubiquitous in clinical care, clinicians need to be vigilant in settings of severe persistent hypolipidemia and/or hypocholesterolemia to evaluate for possible genetic basis for this and to also screen for possible associated complications and comorbidities.

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CARDIOVASCULAR ENDOCRINOLOGY AND LIPIDS DISORDERS CASE REPORT

A Case of Primary Hyperaldosteronism Presenting as Hemorrhagic Stroke

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Background: Primary aldosteronism (PA) is the most common form of secondary hypertension. Patients with PA are more likely to suffer from end-organ damage compared to matched controls with essential hypertension. We present a case of PA identified in a patient who presented with hypertensive emergency and hemorrhagic stroke.
Clinical Case: A 52-year-old man with hypertension and chronic kidney disease presented with sudden onset

left-sided weakness. He had a ten year history of hypertension and was taking carvedilol, losartan, and hydralazine prior to presentation. On arrival, his blood pressure was 263/142 mmHg. He had 3/5 grade weakness in the left upper and lower extremities. Laboratory analysis showed a potassium level of 2.8 mmol/L (n = 3.5–5 mmol/L) and a bicarbonate level of 33 mmol/L (n = 21–29 mmol/L). Screening labs for PA were drawn after potassium repletion. CT Head without contrast revealed an acute 2.5-centimeter intracerebral hemorrhage of the right basal ganglia. He was admitted to the intensive care unit and was started on a nicardipine drip with an improvement of blood pressure. His weakness improved and he was discharged home on carvedilol, hydralazine, nifedipine, and losartan.
Screening for PA revealed a plasma aldosterone concentration (PAC) of 22.8 ng/dL (n < 16 ng/dL) and a plasma renin activity (PRA) of 0.1 ng/ml/hr (n = 0.2–1.6 ng/ml/hr). The PAC/PRA ratio was therefore extremely elevated at 228. The presence of spontaneous hypokalemia, very low renin, and PAC >20 ng/dL confirmed the diagnosis of primary aldosteronism. He underwent an adrenal MRI which revealed two left adrenal nodules, the largest measuring 10 mm, and a 7.3 mm right adrenal nodule, consistent with bilateral adrenal adenomas. The patient did not desire surgery, therefore adrenal vein sampling was deferred. His hypertension improved with the addition of a mineralocorticoid receptor antagonist. Eight weeks after his stroke the patient was readmitted due to chest pain. He was found to have severe multi-vessel coronary artery disease and underwent a four vessel coronary artery bypass.
Conclusion: Patients with PA have higher rates of adverse cardiovascular events compared to age-, sex-, and blood pressure-matched controls with essential hypertension. Studies demonstrate that aldosterone excess has blood pressure independent proinflammatory and profibrotic effects on the vessel wall which leads to endothelial dysfunction and thus accelerated atherosclerosis. Appropriate treatment can eliminate the excess cardiovascular risk associated with PA. This case highlights the importance of including PA in the differential diagnosis of secondary hypertension, particularly among patients presenting with spontaneous hypokalemia, severe uncontrolled hypertension and early onset cardiovascular or cerebrovascular disease.

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CARDIOVASCULAR ENDOCRINOLOGY AND LIPIDS DISORDERS CASE REPORT

Bone Breaking Triglycerides

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A 40 yo African American female with pmhx of T2DM, DLD was admitted for worsening leg and arm pain that started a year prior but had worsened in the last 6 months. Pain started in the right arm and progressed to include the right leg and left leg. She had presented to the ER 3 times in the last 3 weeks with no diagnosis and prescribed anti-inflammatories. On ROS she had unintended weight loss of 50 lbs. Pain was not relieved with anti-inflammatories or narcotics. She was diagnosed with diabetes in the previous 5 years and had not been compliant with her medications. Plain x-rays showed OA of the hip. An osseus survey showed multiple expansile, bubbly, and lucent intramedullary lesions consistent with polyostotic fibrous dysplasia versus multiple myeloma. CT showed a radioluscent lesion of the left femur with absence of normal bone trabeculae. Her labs showed normal calcium, phosphorous, renal function, PTH and no evidence of monoclonal gammopathy. Vitamin D was low at 8.2 ng/ml (6.6–49 ng/ml). CT CAP showed no concern for malignancy in other organs. A lipid profile was done and showed elevated fasting triglycerides of 2617 mg/dL (<150 mg/dl) and LDL direct 54 mg/dl (<100 mg/dl). A1c was 11.2% on admission. She denied any use of alcohol, estrogens, SSRI's. No history of pancreatitis. On physical exam she did not have tendinous xanthomas, eruptive xanthomas, palmar xanthomas, or lipemia retinalis. Family history not significant for lipid disorders. Patient was fasted for 24 hours and then started on intensive insulin regimen as well as fenofibrate for hypertriglyceridemia. Triglycerides came down to less than 500 over 7 days. She was evaluated by ortho for her bone lesions and underwent bone lesion biopsy as well as prophylactic IMN of her bilateral femurs for prevention of impending fragility fractures. Bone biopsy was significant for xanthoma of the bone. Following discharge, she remained on fenofibrate and fish oil as well as a basal/bolus insulin regimen. Triglycerides remained controlled. She has not followed up outpatient for further workup. This case highlights an atypical presentation of triglyceride deposition in the setting of hypertriglyceridemia. It shows that hypertriglyceridemia should be included in the differential for lytic lesions when preliminary workup is negative. It also highlights that complications other than pancreatitis and cardiovascular disease can significantly alter a patient's life if triglycerides go untreated.

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CARDIOVASCULAR ENDOCRINOLOGY AND LIPIDS DISORDERS CASE REPORT

Challenges in Managing Metabolic Complications in a Patient With Familial Partial Lipodystrophy Type 3

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Familial partial lipodystrophy (FPL) is a rare group of autosomal dominant genetic disorders which causes variable loss of subcutaneous fat from abdomen, thorax or extremities in addition to the numerous metabolic complications like insulin resistance, diabetes mellitus and dyslipidemia[1]. FPL type 3 was first characterized by Agarwal et al. in 2002[1], in which peroxisome proliferator-activated receptor-γ (PPARγ) gene was the molecular basis of this disorder. It is extremely rare and so far only 30 patients or so have been recognized with this mutation[2]. FPL3 is unique because it generally spares the loss of fat from trunk, face and neck region and also presents with more severe metabolic derangements. We report a case of a young female