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.

Cardiovascular Endocrinology

CARDIOVASCULAR ENDOCRINOLOGY AND LIPIDS DISORDERS CASE REPORT

Bone Breaking Triglycerides

Ellen Dauterive, MD[1], Virginia Wright, MD[2].
[1]UNIVERSITY OF MISSISSIPPI MEDICAL CENTER, Flowood, MS, USA, [2]University of Mississippi Medical Center, Jackson, MS, USA.

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.

Cardiovascular Endocrinology

CARDIOVASCULAR ENDOCRINOLOGY AND LIPIDS DISORDERS CASE REPORT

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

Kajal Shah, MD[1], Marina Charitou, MD[2].
[1]Stony Brook Medicine/University Hospital Endocrine Fellowship Program, Stony Brook, NY, USA, [2]SUNY Stony Brook, Commack, NY, USA.

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

with PPARγ mutation leading to numerous metabolic complications. A 19 year old female with FPL3 was seen by adult endocrinology as a transition from pediatric endocrinology. She was found to have hypertriglyceridemia on routine labs done at the age of 11. Patient reported loss of subcutaneous fat from her extremities and eruptive xanthoma on flexor surfaces at the time of diagnosis along with a positive family history of hypertriglyceridemia induced pancreatitis and Myocardial infarction at the age of 40 in her father. Her triglyceride level has varied between 600 and 3000 (normal 20–149 mg/dl) over the years. FPL3 was diagnosed based on genetic testing. She was prescribed fenofibrate and fish oil, and statin was added thereafter. She developed type 2 diabetes and was started on metformin and pioglitazone. She was noted to have hypertension and was treated with amlodipine and lisinopril. She also was found to have Polycystic Ovarian Syndrome (PCOS) based on menstrual irregularities, hirsutism and ultrasound showing multiple ovarian cysts, and was treated with spironolactone. Her most recent labs show triglyceride level of 2400 mg/dl and HbA1c of 8.3. PPARγ gene mutation in FPL3 leads to insulin resistance and hence patients often develop hypertriglyceridemia, type 2 diabetes, PCOS and hypertension. In terms of treatment options, we are still limited to pioglitazone, metformin, statins and fish oil. Often these are not sufficient in addressing the complexity of metabolic derangements in these patients who have an increased risk of cardiovascular events at a young age. Further research about agents targeting this gene in particular would be beneficial. 1. Agarwal et al. A novel heterozygous mutation in peroxisome proliferator-activated receptor-gamma gene in a patient with familial partial lipodystrophy. J Clin Endocrinol Metab. 2002 Jan; 87(1):408–411. 2. Garg A. Lipodystrophies: Genetic and Acquired Body Fat Disorders. J Clin Endocrinol Metab. 2011;96(11): 3313–3325.

Cardiovascular Endocrinology

CARDIOVASCULAR ENDOCRINOLOGY AND LIPIDS DISORDERS CASE REPORT

Diet-Responsive Hypercholesterolemia With Cardiofaciocutaneous Syndrome Type 3

Nivedita Patni, MD, Abhimanyu Garg, MD, Chao Xing, PhD.
UT Southwestern Medical Center, Dallas, TX, USA.

Background: Molecular basis of diet responsive hypercholesterolemia remains unclear. We report diet-responsive severe hypercholesterolemia in a young female with cardiofaciocutaneous syndrome type 3 (CFC3) due to a heterozygous pathogenic *MAP2K1* variant, suggesting a role of common MAPK variants in LDL-cholesterol (LDL-C) response to diet. Clinical case: A 3-year-old Caucasian female with CFC3 (macrocephaly, frontal bossing, wide nasal root with depressed bridge, anteverted nares, low set fleshy ears, congenital pulmonic valve stenosis, postnatal growth deficiency, hypotonia, and neurocognitive impairment) due to a *de novo* heterozygous c.389A>G, p.Tyr130Cys pathogenic variant in *MAP2K1,* presented with extremely elevated serum total cholesterol of 446 mg/dL, triglycerides of 239 mg/dL, HDL-cholesterol of 53 mg/dL, LDL-C of 335 mg/dL (normal range < 110 mg/dL) and serum apolipoprotein B level 219 mg/dL (normal range < 90 mg/dL). Her LDL-C was 252 mg/dL a year ago and 215 mg/dL one month prior to presentation. Reducing total dietary fat to 20–25% of total energy and saturated fat to <6% of total energy over the next 4 months lowered LDL-C to 104 mg/dL. However, her weight decreased by 0.5 kg and liberalization of fat intake again increased LDL-C to 222 mg/dL. Her father has mildly elevated LDL-C of 160 mg/dL and her mother had normal LDL-C of 80 mg/dL. Her plasma phytosterol levels were normal and she had ApoE3/E3 genotype. Targeted genetic testing of the patient and parents showed a benign heterozygous LDL receptor (*LDLR*) variant c.2242G>A; p.Asp748Asn, (Minor allele frequency 0.00008) in the patient and her father. Whole exome sequencing of the patient and both parents showed no known disease-causing variants in *LDLR, APOB, PCSK9, LDLRAP1, APOE, STAP1, LIPA, ABCG5, ABCG8* and other known hyperlipidemia-related genes. There are no previous reports of hypercholesterolemia in patients with CFC3. MAP2K1 stimulates various MAP kinases upon wide variety of extra- and intracellular signal and is involved in cell proliferation, differentiation, transcription regulation and development. Previous studies of the relationship between $p42/44^{MAPK}$ activation and *LDLR* expression in human hepatoma HepG2-derived cell line showed that that activation of the Raf-1/MEK/$p42/44^{MAPK}$ cascade induces *LDLR* expression and modulation of the Raf-1 kinase signal strength can determine *LDLR* expression levels. Thus, extent of MAPK activation can alter signaling of LDLR, resulting in hypercholesterolemia. Conclusion: Our case report suggests that MAP2K1 may play a significant role in LDLR signaling, and some MAP2K1 variants may be associated with diet-responsive hypercholesterolemia. Larger studies are required to assess dietary response to LDL-C in subjects with MAP2K1 variants.

Cardiovascular Endocrinology

CARDIOVASCULAR ENDOCRINOLOGY AND LIPIDS DISORDERS CASE REPORT

Familial Partial Lipodystrophy: A Case Study and Review of Recent Literature

Ali Tipu, MD[1], Farhad Hasan, MD[2], Michael Grimes, MD[1].
[1]Allegheny General Hospital, Pittsburgh, PA, USA, [2]Allegheny Health Network, Wexford, PA, USA.

Introduction: Familial partial lipodystrophy (FPLD) is a rare genetic disorder characterized by loss of subcutaneous adipose tissue, mainly from the extremities and gluteal region. FPLD is associated with a variety of metabolic abnormalities including severe hypertriglyceridemia (HTG), insulin resistance (IR), and hepatic steatosis. We present a case of FPLD and summarize recent literature on the metabolic features and their management in patients with this rare disease. Case: A 44 year old female with medical history of Type 2 DM, hypertension, hypothyroidism and recurrent pancreatitis from severe HTG was referred to our clinic. She was diagnosed with Type 2 DM in her 30s. Over the ensuing years she had significant IR requiring increasing doses of concentrated insulin (up to 250 units/day). She reported progressive loss of subcutaneous fat from extremities in the preceding 2–3 years. She