Each patient's consent was obtained over the phone or during a visit to the clinic. Primary data were collected with a questionnaire, whereas additional data, including HbA1c levels before and after starting CSII, were collected from medical records. A total of 57 participants were enrolled in the study. African Americans represented 79% of the participants; 43% of the participants were unemployed, and 56% had an annual income of less than 20,000 USD. Since commencing CSII therapy, all participants achieved a decrease in mean HbA1c level from 9.7% to 8.0% (P = 0.001), and that of African American participants decreased from 9.8% to 8.2%. Increase number of individuals at home was associated with less reduction in HbA1c levels after starting CSII therapy (P = 0.02). Overall, satisfaction with CSII therapy was high, and 63% of participants reported being very satisfied with the treatment. The mean BMI among participants while using MDI was 32.6 kg/m^2 but significantly increased to 33.9 kg/m^2 (P = 0.01) while using CSII. The increase in mean BMI after starting CSII therapy was significantly higher in participants with T2D than in ones with T1D (P = 0.001). While receiving MDI, female participants had a significantly higher mean BMI than their male counterparts (P = 0.02); however, that difference became nonsignificant after they began CSII therapy (P = 0.06). The level of physical activity after starting CSII therapy did not alter the risk of increased BMI. The results of our interim analysis indicate the significant effect of CSII in lowering HbA1c levels in all diabetic patients regardless of sex, race, BMI, type of diabetes, marital status, employment status, level of education, adherence to diabetic diet, physical activity, duration on CSII, and use of other antidiabetic medications. The significant increase in BMI once CSII therapy commenced may reflect the increase in insulin dose among patients who were not adherent to insulin while receiving MDI. Patients need to be aware of that side effect, and additional interventions for weight management may be considered for overweight and obese patients planning to start treatment with CSII.

Cardiovascular Endocrinology

HYPERTRIGLYCERIDEMIA; INFLAMMATION AND MUSCLE METABOLISM IN OBESITY AND WEIGHT LOSS I

Familial Homozygous Lipoprotein Lipase Defect Presenting with Recurrent Chylomicronemia Syndrome: Making a Case for Elective Plasmapheresis as an Adjuvant Treatment Modality.

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SAT-565

Background

The chylomicronemia syndrome is a disorder characterized by severe hypertriglyceridemia and fasting chylomicronemia. Type Ia hyperlipoproteinemia is an extremely rare genetic disorder that results from homozygous deficiency in LPL activity. It is characterized by eruptive xanthomas, lipemia retinalis, memory disturbances, hepatosplenomegaly and frequent episodes of pancreatitis. Pharmacologic agents including fibrates, fish oils and statins have been used for treatment. Patients who fail pharmacologic therapy are usually treated with plasmapheresis. This case showed a patient in which joint decision making led to elective plasmapheresis to avoid chylomicronemia syndrome as an adjunct to medical therapy.

Clinical case

A 35-year-old lady without known medical co-morbidity presented with 2 weeks of dull abdominal pain radiating to the back. She developed nausea and vomiting which led to her presentation. There was no history of alcohol use, gall stones, diabetes mellitus or thyroid dysfunction. She was not on any medications. Physical examination was significant for BMI of 18, moderate abdominal tenderness, splenomegaly and an indurated rash in her legs. Laboratory investigation showed elevated lipase and triglyceride level of 3313 (normal 30-50) mg/dL. CT of the abdomen was consistent with acute interstitial pancreatitis. She was managed with insulin drip and fenofibrate and discharged 3 days later on fenofibrate, atorvastatin and long acting insulin. She developed another episode of acute pancreatitis while on medical therapy requiring readmission and initiation of insulin drip 2 months later. However, triglycerides trended upwards when the drip was stopped and symptoms of acute pancreatitis worsened. She subsequently underwent plasmapheresis which led to resolution of symptoms. Despite maximally tolerated pharmacologic therapy, she persistently has triglycerides above 4000s and persistent abdominal discomfort. Genetic testing confirmed homozygous defect in LPL gene. Following an outpatient Endocrinology visit, a decision was made to pursue elective plasmapheresis as an adjunct to therapy. She had on average 2 sessions monthly for 3 months with overall improvement in abdominal discomfort as well as significant improvement in triglyceride levels.

Conclusion

Familial chylomicronemia syndromes often require multimodal therapeutic approaches to prevent morbidity and complications. These include diet and pharmacologic therapy. Although plasmapheresis is often used during hospitalizations for hypertriglyceridemia induced pancreatitis refractory to diet and pharmacologic therapy, it was used in our patient electively and efficaciously to control hypertriglyceridemia and improve symptoms of chylomicronemia syndrome.

Reproductive Endocrinology

SEX, GENDER, AND HORMONES

Combining Clinical and Genetic Approaches in Diagnosing a Large Brazilian Cohort of Patients with 46,XY Differences/Disorders of Sex Development (DSD)

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OR27-01

Background: It is recommended a multidisciplinary approach consisted of clinical, hormonal and genetic workups for diagnosing 46,XY DSD. However, no previous study has quantified how useful is this combined approach.

Objectives: To retrospectively review the clinical and genetic findings for diagnosing a large cohort of patients with 46,XY DSD from a single Brazilian center.

Methods: 247 non-syndromic 46,XY DSD individuals (159 sporadic and 88 familial cases from 39 families) were studied. Clinical and hormonal data were collected from medical files. Testosterone (T), androstenedione (A) were measured by immunoradiometric or immunofluorimetric assays and dihydrotestosterone (DHT) by RIA after celite chromatography or by liquid chromatography tandem mass spectrometry; T/DHT and T/A ratios were calculated. Analysis of sensitivity (SE), specificity (SP) of T/DHT was performed, being the molecular diagnosis considered the gold standard for diagnosing SRD5A2 deficiency. A T/A>0.8 was considered indicative of 17ß-HSDB3 deficiency. The patients were clinically classified into four subgroups: 1) androgen insensitivity syndrome (AIS), 2) gonadal dysgenesis (GD); 3) defects in androgen synthesis (DAS) and 4) DSD of unknown etiology. Molecular studies were performed by Sanger sequencing and/ or massively parallel sequencing (MPS).

Results: The median age at first visit was 14 years (range 0.1 to 59 years). The molecular diagnosis was established in 96.5% of the cases with AIS (n=28/29), in 96% of the subjects with DAS (n=46/48), in 36% of the patients with GD (n=21/57) and in 26.7% (n=15/56) with DSD of unknown etiology. The best cut-off for T/DHT in basal state and hCG stimulated was 12.5 (SE=100%; SP=78.57%) and 24 (SE=87.5%; SP=95.7%) respectively. A T/A<0.8 was observed in 13/16 (81%) of the patients with molecular diagnosis of 17ß-HSDB3 deficiency and also in 1/49 patients with other diagnose. Classification according to the phenotype matched with the genetic diagnosis in most cases. The molecular evaluation allowed that 16% (9/56) of the patients that were classified as DSD of unknow etiology had a definitive diagnosis, including six GD cases, two individuals with SRD5A2 deficiency and one with 17ß-HSDB3 deficiency. A clear AIS phenotype of five patients allowed us to consider and prove the pathogenicity of two synonymous and one promoter region variants as the cause of AIS. The combination of clinical and molecular diagnosis led to an increase in 8% the diagnosis in a total of 116 index-cases (58.5%) with a molecular diagnosis.

Conclusion: Considering the phenotype heterogeneity, pitfalls of the hormonal assessment and number of genes involved, it is reasonable to consider MPS as a first test for diagnosing patients with 46,XY DSD. However, the combination of clinical and molecular diagnosis is more accurate than either strategies alone in diagnosing 46,XY DSD.

Adipose Tissue, Appetite, and Obesity

RARE CAUSES AND CONDITIONS OF OBESITY: PRADER WILLI SYNDROME, LIPODYSTROPHY

A Rare Presentation of Early Onset Wernicke Encephalopathy Following Sleeve Gastrectomy

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SUN-596

A Rare Presentation of Early Onset Wernicke Encephalopathy Following Sleeve Gastrectomy

Background: Wernicke encephalopathy (WE) has been reported after malabsorptive bariatric surgeries but is an uncommon complication of sleeve gastrectomy. Since 2011, the number of patients receiving a sleeve gastrectomy has tripled, with almost 60% of patients undergoing bariatric surgery receiving a sleeve gastrectomy in 2017 (1). We present a case of WE in a young woman as a rare and early complication of sleeve gastrectomy.

Clinical Case: A 32-year old female with a past medical history significant for hypertension,
pseudotumor cerebri, and morbid obesity status post sleeve gastrectomy two months prior presented to the emergency department with complaints of blurry vision and lower extremity numbness. Physical examination showed sluggish light reflex and decreased extraocular movements. Given history of pseudotumor cerebri, patient underwent a therapeutic lumbar puncture with removal of 13 ml of CSF. Opening pressure was 20 cm of water and patient experienced no relief of her symptoms. Ophthalmology consult did not offer an explanation for the blurry vision. MRI-brain with and without contrast showed findings highly suggestive of
WE. It showed faint linear symmetric hyperintensities along the bilateral mesial thalamus, dorsal midbrain and periaqueductal gray matter, which were determined to be acute in nature in comparison with a MRI performed three weeks prior. Upon further investigation, thiamine level was low at 43.6 nmol/L [66.5 – 200] confirming the diagnosis of WE. Thiamine supplementation was started immediately and patient reported improvement of her vision the next day with return to baseline in 3 days.

Conclusion: There have been a handful of cases of WE reported in literature as a complication of sleeve gastrectomy. Zheng, L also reported a case of WE 7 weeks after sleeve gastrectomy (2). Although sleeve gastrectomy does not directly affect the primary absorptive pathway of thiamine in the gastrointestinal tract, it is imperative to consider WE in patients presenting with suspicious neurologic symptoms after a recent sleeve gastrectomy. WE was suspected in our case due to typical MRI findings and neurological presentation after bariatric surgery, which was later confirmed by low serum thiamine level. Early detection and thiamine supplementation resulted in complete reversal of symptoms in our patient. WE is a rare but severe and preventable consequence of bariatric surgery that warrants attention given its rapid onset and detrimental course.