

MON-LB120

Background: Maturity onset diabetes in young 3 (MODY 3) is caused by mutation of the hepatic nuclear factor 1 alpha (HNF-1A) gene. Craniofacial macrosomia (CFM) is associated with an abnormal development of craniofacial structures during the embryonic period. Maternal diabetes and genetic predisposition have been associated with CFM¹. There are rare reports about an association of MODY 3 and CFM. **Clinical case:** An 11-year-old male patient presented with right side CFM (mild mandibular hypoplasia, internal auditory canal absence, severe pinna hypoplasia, abnormal orbital size and location, O3.M0.E3.N0.S0²) noted at 8 months of age. Preoperative examination revealed A1c at 10.9%. After short term intensive insulin therapy, he had standard bread meal test: fasting glucose 8.11 mmol/L, insulin 13.9 mIU/L (3-25), C-peptide 1.25 ng/ml (0.81-3.85); 1 hour glucose 10.05 mmol/L, insulin 27 mIU/L, C-peptide 2.42 ng/ml; 2 hour glucose 8.17 mmol/L, insulin 16.09 mIU/L, C-peptide 2.11 ng/ml. GADA, IAA and ICA were negative. The mother was diagnosed diabetes at age 27years, when the patient was 8-month-old, and received insulin therapy. The mother was blind by age 35years due to diabetic retinopathy and died of DKA at 38-years-old. The patient's 16-year-old brother had left side CFM (O2.M1.E2.N0.S0) and his OGTT was normal. The father was diagnosed with impaired glucose tolerance. The family had whole genome sequencing by Sanger technique, and resequenced the mutation with side primers. The CGA to CAA mutation was present at the 686 loci of exon 3 of HNF1A gene in the patient and mother. The HNF1A exon 3 mutation of CGA to CAA resulted in the change of arginine to glutamine which by the HGMA database is recognized as a reported MODY3 gene mutation. There was a mutation of G to A in the 4 loci of exon 1 of the insulin coding region in chromosome 11 in both the patient and elder brother. Neither elder brother nor father had the CGA mutation of HNF1A. **Conclusion:** There has not been a previous report of a relationship between HNF1A and CFM. In this case, the elder brother had CFM without a HNF1A mutation which does not support a connection between CFM and HNF1A. The two brothers both had CFM and insulin coding gene mutations which would represent a new association not previously described. Further testing is needed to confirm a relationship between the two. **Reference:** 1. Chen Q, Zhao Y, Shen G, Dai J. Etiology and Pathogenesis of Hemifacial Microsomia. *J Dent Res* 2018; 97(12): 1297-305. 2. Gougoutas AJ, Singh DJ, Low DW, Bartlett SP. Hemifacial microsomia: clinical features and pictographic representations of the OMENS classification system. *Plast Reconstr Surg* 2007; 120(7): 112e-20e.

Cardiovascular Endocrinology**HYPERTRIGLYCERIDEMIA; INFLAMMATION AND MUSCLE METABOLISM IN OBESITY AND WEIGHT LOSS I****Rapid Reduction of Insulin Requirement in a Hyperglycemic Patient Treated for Hemophagocytic Lymphohistiocytosis**

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SAT-LB93

Background: Hemophagocytic lymphohistiocytosis (HLH) is life-threatening disorder of immune dysregulation involving macrophage and T-cell activation resulting in massive cytokine release causing multi-organ dysfunction. Similar release of cytokine products from fat tissue is associated with obesity-related insulin resistance. Our case presentation is an example of HLH and insulin resistance, two conditions with overlapping pathophysiology, occurring simultaneously. **Clinical Presentation:** A 17-year-old male, with no history of hyperglycemia, underwent renal transplant due dysplastic kidneys. He received 500mg IV methylprednisolone during surgery followed by a prednisone taper starting at 70mg daily. Serum glucoses post-transplant ranged from 97 to 129 mg/dL. Three weeks post-transplant he was admitted for fever and dehydration. BMI on admission was the 85th percentile. Serum glucose was 371 mg/dL without ketosis. He started on insulin therapy, requiring 60 units per day (0.8 units/kg). It was suspected his new-onset diabetes was due to his immunosuppressant regimen (prednisone 50mg daily, tacrolimus) and/or acute illness. With persistent fevers and negative infectious workup, there was concern for HLH. The diagnosis was confirmed with ferritin level of 65,962 ng/mL (27-265), hemoglobin 6.5 gm/dL, platelets 88,000, triglycerides 765 mg/dL, soluble IL-2 receptor 2,717 u/mL (45 - 1,105). For HLH treatment, he received methylprednisolone 800 mg daily x 3 doses. During this time his insulin requirements increased to 188 units per day (3.6 units/kg). He was transitioned to dexamethasone 20mg daily. His insulin requirements increased over the next 72 hours to 388 units per day (5.2 units/kg). He was found to be positive for Ehrlichiosis, a known precipitant of HLH. Doxycycline therapy was initiated for a 14 day course. One week into his doxycycline course his ferritin had decreased to 999 ng/mL. He remained on dexamethasone 20 mg daily but developed severe hypoglycemia to 29 mg/dL with altered mental status. All insulin therapy was held. Fasting glucoses over the next 4 days ranged from 94-154 mg/dL and post-prandial glucoses 116-288 mg/dL. He discharged home with only short acting insulin for glucoses above 250 mg/dL, which he did not require. **Case Lessons:** Cytokine release from macrophages is implicated in the pathology of both HLH and insulin resistance associated with obesity. Glucocorticoids used to treat HLH can also exacerbate insulin resistance. Providers should be aware of the risk of hyperglycemia and large insulin requirements in patients with HLH, and the potential for rapid reduction of insulin needs as HLH is successfully treated.

Healthcare Delivery and Education**EXPANDING CLINICAL CONSIDERATIONS FOR PATIENT TESTING AND CARE****Studying the Care and Social Pathway of Young Adults With Endocrine and Metabolic Diseases During Transition: The "Transend" Cohort**

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MON-LB308

Context. The transition period between pediatric and adult medicine is associated with poor patient outcomes and an important number of patients lost to follow up. Intervention exist but the few published randomized trials do not allow to study long-term patient outcomes nor intervention sustainability in time. **Objective.** Describe the cohort of patients in adult care who benefit from a new transition program based on case management approach, its activity and follow-up outcomes. **Methods.** A longitudinal study was led since September 2016 in adult services of endocrinology, nutrition and diabetology of a French University Hospital. Patients with any endocrine disease diagnosed during childhood and transferred to adult care were included. The care pathway for these patients was built in three steps. Step 1 is dedicated in liaising with pediatric services and patient to facilitate its first visit in adult care. Step 2 defines the care pathway in adult service based on the needs assessment realized by the coordinator upon the patient's arrival in adult service. Step 3 focuses in liaising with structures outside hospital (GP, educational and social sector). Thorough the follow-up, the coordinator is identified as the key contact by the patients. Attendance to medical appointments, clinical, and social data are collected throughout patient follow-up. **Results.** Since 3 years, 500 patients benefited from the case management mainly for their obesity (n=91, 18%), type 1 diabetes (n=54, 11%), malignant brain tumor (n=68, 14%) or congenital hypopituitarism (n=42, 8%). They were aged 19 in median at transfer in adult care, sex ratio: 0,5, A large majority live in the parental home (409, 82%), 169 (34%) are university students, 130 (26%) are in high school, 90 (18%) are in medico-social institution. Patients who required most of support from the coordinator usually combine one (or more) somatic disease and either a neuro-cognitive disorder or a psychiatric disorder, they all have social difficulties. In patients with more than 3 months of follow-up (median: 18 months), 22/418 (5%) are out of follow-up. Concerning the patients for whom the follow-up is 36 months or more, the percentage of out of follow-up is the same: 5% **Conclusions.** The case manager addresses the complex needs of diverse patients. With time, the cohort will provide unprecedented long-term results of patients with various conditions who went through transition.

Reproductive Endocrinology

TRANSGENDER MEDICINE AND RESEARCH

Cross Sex Hormone Therapy and Breast Cancer in Transgender Male to Female

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SUN-LB8

Case Presentation: We are presenting a 53-year-old Male to Female transgender patient who has been receiving estradiol valerate injections every 14 days for 13 years and had no gender surgical reassignment procedures or breast implants. Her past medical history was significant for HIV on Highly Active Anti-retroviral therapy (HAART). No family history of breast cancer. She presented with severe bilateral left elbow, lower back and bilateral chest pain for 3 days. Chest CT done to exclude pulmonary embolism showed an incidental 4 cm right breast mass. Enlarged lymph nodes in the right axilla, scattered lytic lesions in the axial skeleton and the left humeral head were also noted. Breast exam was not performed until the significant findings were seen in CT chest and it showed a palpable hard-circumscribed subareolar right breast mass without skin changes. Ultrasound guided biopsy of the breast mass confirmed invasive ductal carcinoma of the breast. The patient had no previous mammogram testing. Oncology work-up was positive for estrogen and progesterone receptors but negative for human epidermal growth factor-2 receptor. The patient opted to return home in another state to seek treatment and further oncological workup but subsequently lost follow up. **Discussion:** Male to female breast cancer was first recognized in 1968. However, risk factors for this condition remain unclear. In our patient, long-term use of Cross-sex Hormone Therapy (CHT) represented a major risk factor for breast cancer. In a Dutch study, the risk of breast cancer increased during a relatively short duration of CHT and the cancer characteristics reassembled female pattern. As theoretically implicated, increased estrogen exposure in males may have a role in the proliferation of neoplastic breast epithelium. There are growing evidence to support increasing rates of breast cancer in HIV-positive population, making it a potential risk factor as well. Loss of CXCR-4 protective effect promoted by HIV virus may explain the increase in the breast cancer incidence after the introduction of HAART. In general, routine screening for breast cancer in MTF transgender population remains controversial. The Endocrine Society Clinical Practice guidelines suggest that MTF transsexual individuals who have no known increased risk of breast cancer should follow screening guidelines for biological women. While the Canadian Cancer Society recommends screening mammography every two years for MTF individuals taking CHT for more than 5 years and those between the ages of 50 and 69 years.

Conclusion: Breast cancer in MTF transgender patients is associated with receiving CHT and represents diagnostic and treatment challenge. More research is need to comment on routine breast cancer screening in this population. However, physicians should remember performing a regular breast exam in MTF individuals looking for a possible mass.

Thyroid

THYROID DISORDERS CASE REPORTS IV

Thyrotoxic Periodic Paralysis in Hispanic Patients

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