association between the rare p.L46P substitution and dyslipidemia. The purpose of this study is to analyze the lipid profile of patients with the APOE c.T137C:p.L46P missense mutation to identify an association with a specific phenotype.
Methods: Next generation sequencing was performed on patients seen in the Lipid Genetics Clinic in London, Ontario, since 1998. Patients heterozygous for the APOE c.T137C:p.L46P mutation were identified. Untreated lipid profiles were imputed and compared with healthy controls.
Results: A total of 9 patients met the inclusion/exclusion criteria. Mean total cholesterol, triglycerides, HDL-C and LDL-C in mutation-positive subjects were 6.83±1.84, 2.91±1.67, 1.03±0.28 and 4.30±1.90 mmol/L, respectively. In healthy controls, the mean values were 4.81±0.82, 1.05±0.37, 1.27±0.41 and 3.18±0.83 mmol/L, respectively ($P<0.05$ for all except HDL-C). The p.L46P allele frequency in patients with dyslipidemia was 0.74% compared with 0.25% in the general population (odds ratio, 2.96; 95% CI, 2.06 to 4.24; $P<0.05$). 55.6% of mutation-positive individuals had clinical evidence of coronary artery disease with an average age of onset of 62.2±12.9 years.
Conclusions: Individuals with the APOE c.T137C:p.L46P missense mutation were found to have elevated LDL-C and elevated triglycerides, suggesting an association with combined hyperlipidemia. Patients referred with dyslipidemia were significantly more likely to have this mutation compared with the general population. This rare mutation can be considered in diagnostic genetic screening in patients with hypertriglyceridemia and hypercholesterolemia.

112

Assessing Quality of Thyroid Referral Requests from Primary Care Providers to Endocrinologists in Ottawa

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Background: Referral requests from primary care providers (PCPs) to endocrinologists may not always include essential data required for effective triaging and initial consultation. The purpose of this project was to assess the quality of faxed thyroid referrals from PCPs and determine whether essential information was included.
Methods: Two coders (G.G. and J.W.) reviewed faxed thyroid referrals from PCPs for components of the QuRE checklist (Alberta Health Services), and essential information based on clinical practice and Choosing Wisely Canada guidelines. Descriptive statistics were used to describe findings.
Results: Of 261 thyroid referrals (including hypothyroidism, hyperthyroidism, thyroid nodule(s), Graves' disease, goiter, thyroiditis and thyroid cancer) received in 2020, 140 were from PCPs. Average length of referral was 6.5 pages (range: 1-32 pages) for hyperthyroidism (N=46) and 7.5 pages (range: 3-15 pages) for thyroid nodules (N=21). Hyperthyroidism referrals included TSH 91%, free T4 76% and free T3 59% of the time, respectively. Ninety-five percent of thyroid nodule referrals included an ultrasound and 76% included TSH. None of the faxed hyperthyroidism or thyroid nodule referrals met all of the QuRE checklist criteria, with 57% of hyperthyroidism and 62% of thyroid nodule referrals missing comments about urgency.
Conclusion: Hyperthyroidism and thyroid nodule referral requests vary in length and information included, thus affecting triaging for urgency and appropriateness. Locally, as we move towards an eReferral system, it will be worthwhile to develop a standard referral template to help PCPs build high-quality referrals.

113

Detecting Early Markers of Cardiovascular Disease in High-Risk Women With and Without PCOS

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Background: Women with polycystic ovary syndrome (PCOS) are at increased risk of cardiovascular disease (CVD). Currently, there is a need screen for early CVD in high-risk women with and without PCOS to effectively assess, manage, and prevent morbidity from CVD.
Objective: This study aims to provide evidence-based research that will aid assessment guidelines for early detection of dyslipidemia, cardiac dysfunction and subclinical atherosclerosis in women with and without PCOS.
Design, setting, participants: A case-control study in high cardiometabolic risk women aged 25-45 years with and without PCOS matched for age and body mass index.
Main outcome measures: ACVD is measured using carotid intima-media thickness (cIMT) and cardiac function using ultrasound and 2D/3D echocardiography. Plasma insulin, glucose, apo-B lipoproteins, triglycerides (TGs), LDL-cholesterol (C), HDL-C, and non-HDL-C.
Results: Preliminary data (n=14 PCOS, and n=2 control) shows PCOS women have decreased Left ventricular (LV) ejection fraction (EF) (59.86 ± 2.48) compared to controls (64 ± 1.41), indicating early systolic dysfunction. Those with PCOS tend to have increased LV mass (24.4%), cIMT (19.9%), and impaired LV Global Longitudinal Strain (GLS) (1.4%) compared to those without PCOS. No significant difference in fasting plasma lipids and apoB- lipoproteins are observed in controls and PCOS.
Conclusion: Our preliminary data shows no significant differences in fasting atherogenic dyslipidemia, and women with PCOS have early impairment in systolic and global LV function compared to controls.

114

Testosterone Suppression With Injectable Estrogen Therapy Alone in Male to Female Transgender Patients

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Background: Hormone therapy for male to female (MTF) transgender patients aims to achieve testosterone suppression and estrogen augmentation with target hormone levels in the cis-female range. Current guidelines recommend concurrent use of anti-androgens and estrogen formulations to achieve this goal. There is limited data to confirm that both agents are needed in all patients.
Methods: A retrospective analysis of testosterone levels and anti-androgen therapy in 190 MTF patients presenting to West Ottawa Specialty care between July 1, 2014, and July 2019 was conducted.
Results: Three patients in our cohort with testes in situ requested to be on injectable estrogen alone without anti-androgen therapy. All had baseline cis-mail range testosterone levels and received guideline based estradiol doses. Subsequent testosterone levels were suppressed to the target range.
Conclusions: Injectable estradiol therapy alone in MTF patients with testes in situ is sufficient to achieve target testosterone levels. Future studies are needed to evaluate the safety of this approach.