10th International Meeting of Pediatric Endocrinology: Individual Abstracts for Free Communication and Poster Sessions

FREE COMMUNICATION SESSIONS:

Friday, September 15, 2017, 7:30-8:30am

Growth and GH/IGF Axis #1: FC1 – FC5
Syndromes: FC6 – FC 10
Bone and mineral metabolism #1: FC11 – FC15

Friday, September 15, 2017, 8:45-9:45am

Sex differentiation/gonads and disorders of sex development: FC16 – FC20
Type 2 diabetes and other carbohydrate metabolism: FC21 – FC25
Global health: FC26 – FC30

Friday, September 15, 2017, 2:30-3:30pm

Bone and mineral metabolism #2: FC31 – FC35
Type 1 diabetes #1: FC36 – FC40
Gender dysphoria: FC41 – FC45
Obesity, lipids, and co-morbidities #1: FC46 – FC50

Saturday, September 16, 2017, 7:30-8:30am

Quality improvement: FC51 – FC55
Growth and GH/IGF Axis #2: FC56 – FC60
Obesity, lipids, and co-morbidities #2: FC61 – FC65

Saturday, September 16, 2017, 8:45-9:45am

Type 2 diabetes and other carbohydrate metabolism #2: FC66 – FC70
Thyroid: FC71 – FC75
Neuroendocrinology including hypothalmic pituitary: FC76 – FC80

Saturday, September 16, 2017, 3:15-4:15pm

Type 1 diabetes #2: FC81 – FC85
Fetal and neonatal endocrinology and metabolism, including hypoglycemia: FC86 – FC90
Adrenals #1: FC91 – FC95

Sunday, September 17, 2017, 7:30-8:30am

Adrenals #2: FC96 – FC100
Multisystem endocrine disorders: FC101 – FC105

Sunday, September 17, 2017, 8:45-9:45am

Puberty: FC106 – FC110
Late Breaking: FC111– FC115

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POSTER SESSION 1

Thursday, September 14, 2017, 5:45-6:45pm

P1 – Adrenals: P1-100 – P1-135
P1 - Bone and mineral metabolism: P1-200 – P1-234
P1 - Fetal and neonatal endocrinology and metabolism, including hypoglycemia: P1-500 – P1-526
P1 - Growth and GH/IGF Axis: P1-800 – P1-860
P1 - Multisystem endocrine disorders: P1-900 – P1-915
P1 - Neuroendocrinology including hypothalmic pituitary: P1-1000 – P1-1016
P1 - Obesity, lipids, and co-morbidities: P1-1100 – P1-1136
P1 – Other: P1-1200 – P1-1216
P1 – Puberty: P1-1300 – P1-1321
P1 - Quality improvement: P1-1400 – P1-1413
P1 - Sex differentiation/gonads and disorders of sex development: P1-1500 – P1-1522
P1 – Syndromes: P1-1600 – P1-1615
P1 – Thyroid: P1-1700 – P1-1727
P1 - Type 1 diabetes: P1-1800 – P1-1828
P1 - Type 2 diabetes and other carbohydrate metabolism: P1-1900 – P1-1907

POSTER SESSION 2

Friday, September 15, 2017, 11:30am-12:30pm

P2 – Adrenals: P2-100 – P2-130
P2 - Bone and mineral metabolism: P2-200 – P2-219
P2 - Ethics in endocrinology: P2-400 – P2-401
P2 - Fetal and neonatal endocrinology and metabolism, including hypoglycemia: P2-500 – P2-530
P2 - Global health: P2-700 – P2-704
P2 - Growth and GH/IGF Axis: P2-800 – P2-857
P2 - Neuroendocrinology including hypothalmic pituitary: P2-1000 – P2-1014
P2 - Obesity, lipids, and co-morbidities: P2-1100 – P2-1146
P2 – Other: P2-1200 – P2-1208
P2 – Puberty: P2-1300 – P2-1326
P2 - Sex differentiation/gonads and disorders of sex development: P2-1500 – P2-1533
P2 – Syndromes: P2-1600 – P2-1620
P2 – Thyroid: P2-1700 – P2-1734
P2 - Type 1 diabetes: P2-1800 – P2-1852
P2 - Type 2 diabetes and other carbohydrate metabolism: P2-1900 – P2-1906

POSTER SESSION 3

Saturday, September 16, 2017, 12:00-1:00pm

P3 – Adrenals: P3-100 – P3-132
P3 - Bone and mineral metabolism: P3-200 – P3-238
P3 - Endocrine care transition: P3-300 – P3-304
P3 - Fetal and neonatal endocrinology and metabolism, including hypoglycemia: P3-500 – P3-525
P3 - Gender dysphoria: P3-600 – P3-608
P3 - Growth and GH/IGF Axis: P3-800 – P3-863
P3 - Neuroendocrinology including hypothalmic pituitary: P3-1000 – P3-1012
P3 - Obesity, lipids, and co-morbidities: P3-1100 – P3-1142
P3 – Puberty: P3-1300 – P3-1331
P3 - Sex differentiation/gonads and disorders of sex development: P3-1500 – P3-1539
P3 – Syndromes: P3-1600 – P3-1619
P3 – Thyroid: P3-1700 – P3-1741
P3 - Type 1 diabetes: P3-1800 – P3-1849
P3 - Type 2 diabetes and other carbohydrate metabolism: P3-1900 – P3-1911

1.8mmol/L) confirming the diagnosis of CHI. The hypoglycaemia was unresponsive to maximum dose of diazoxide (20mg/kg/day) and she was subsequently managed with a combination of octreotide (40mcg/kg/day) and sirolimus therapy
Results: Genetic analysis revealed a homozygous nonsense mutation in *ABCC8* (c.1990C>T, p.Gln664Ter) inherited from both parents. The urinary analysis showed a persistently elevated glycolate (>1000µmol/L) and oxalate (>800 µmol/L) levels giving rise to the clinical suspicion of primary hyperoxaluria type 1 (PH1). However, further genetic analysis did not show mutation in *AGXT* but revealed a homozygous missense mutation in *HAO1* (c.493G>T, p.Gly165Cys). Enzyme activity analysis on liver biopsy revealed a normal AGT enzyme activity but absent GO enzyme activity, confirming GO deficiency.
Conclusions: This is the first reported combination of two rare autosomal recessive conditions namely CHI and GO deficiency in a patient with unexplained hyperoxaluria. GO deficiency is not predicted to have a pathological phenotype but the unusual association of hyperoxaluria can cause diagnostic difficulties and warrants a careful monitoring of renal function in the long-term.

POSTER SESSION 3
Saturday, September 16, 2017, 12:00-1:00pm
P3 - Gender dysphoria
P3-600 – P3-608

P3-600

GUIDELINE-DRIVEN LABORATORY TESTING IN THE CARE OF TRANSGENDER YOUTH: ARE ALL THE RECOMMENDED TESTS REALLY NEEDED?

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Objectives: Published guidelines addressing the care of transgender youth call for aquisition of many baseline and post-treatment laboratory tests. The aim of this project was to assess the utility of this routine testing.
Methods: Charts from 203 transgender adolescents were reviewed to obtain baseline demographic data and results of blood tests. Baseline blood tests were performed at the initial visit and included hormonal, renal, hepatic and hematologic tests ; suppression labs (LH, FSH, testosterone, estradiol) were performed while on leuprolide acetate (Lupron depot) ; and safety labs (repeat of baseline testing) were performed while on cross-sex hormones to evaluate sex steroid levels and screen for potential complications. Data were analyzed with descriptive statistics, and paired t-tests were used for comparing baseline and follow-up tests.
Results: 156 and 47 male bodied youth (MBY) were included. Mean age at presentation was 16.3 years SD 1.63 for FBY and 16.1 years SD 1.70 for MBY. Mean bone age was 15.9 years in both FBY (SD 1.3, range 12-18) and MBY (SD 2.2, range 11-19). In 9 FBY (7.7%) and 2 MBY (6.5%), bone age was advanced more than 2 SD. Mean Tanner stage was 4.4 SD .8 for FBY (B4P4, median 5, mode 5) and 4.0 SD 1.1 for MBY (G4P4, median 4, mode 5). Baseline blood tests showed no abnormal hormonal results except in one youth with elevated FSH who was subsequently found to have Klinefelter syndrome. In 6 (4.5%) FBY, testosterone levels were slightly high (max value: 2.5 nmol/L) according to sex and Tanner stage norms. Suppression blood tests showed adequate suppression for LH and FSH in all youth. Safety tests did not reveal the development of any clinically significant abnormalities while on sex hormone therapy. However, statistically significant differences in hemoglobin levels (increased in FBY on testosterone p=.002, decreased in MBY on estradiol p=.019) and in red blood cell count (p=0.000; increased in FBY and decreased in MBY) were observed.
Conclusions: Our results need to be corroborated by additional studies. However, the results suggest that much of the routine testing recommended in care guidelines may not be warranted. Reducing unnecessary testing could improve both care for transgender youth and resource utilization.

P3-601

USE OF FERTILITY PRESERVATION BY TRANSGIRLS STARTING HORMONAL TREATMENT

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Objectives: Many transgender adolescents undergo treatment with GnRH analogues, followed by cross sex hormones and, in adulthood, gender affirming surgery which may include gonadectomy. Infertility as a consequence of such treatment may have a major impact on their lives. Current guidelines recommend fertility counselling. We aimed to investigate the use of fertility preservation in a Dutch cohort of transgirls.
Methods: Data on counselling about fertility preservation and its actual use were extracted from the medical files of transgirls who started treatment with a GnRH analogue between 2011 and 2016.
Results: The possibility of semen cryopreservation was discussed with 27/28 transgirls. Eleven adolescents were referred for semen cryopreservation, one of whom was unable to produce a semen sample. Three declined because they did not want to have children; two because they wanted to adopt; one because she abhorred the procedure and thought she would not use her semen; one because she disliked the idea of a women being impregnated with her semen. In five no reason for declining fertility preservation was noted. Four individuals were in early puberty with testicular volume ≤8 ml; because semen cryopreservation was unlikely to be successful they were not referred. The one individual to whom semen cryopreservation was not offered had a testicular volume of 4-5 ml so was also unlikely to be able to produce a semen sample through masturbation.

average BMI z-scores between male-to-female (M2F) and female-to-male (F2M) transgender patients.
Methods: A retrospective review of patients with GD followed in the pediatric endocrine clinic at Riley Hospital for Children was performed. Variables analyzed included age, natal sex, affirmed gender, weight and height at the first clinic visit. BMI percentiles and z-scores were calculated based on height and weight. An independent samples t-test was conducted to compare average BMI z-scores for M2F and F2M patients.
Results: Seventy-eight patients with GD were identified of whom 29 (37%) were M2F and 49 (63%) were F2M. Within the M2F cohort, aged 14.2±3.15 years, 86.2% were Caucasian and average BMI z-score was 0.47±1.51. Based on BMI percentile, 7 (24.1%) patients were overweight and 7 (24.1%) were obese. In the F2M cohort, aged 14.6±2.0 years, 85.7% were Caucasian and average BMI z-score was 0.87±1.15. Ten (20.4%) patients were overweight and 15 (30.6%) were obese. In the group as a whole, five patients (6.4%) were underweight and the remaining were of normal weight. No difference was seen in average BMI z-scores between M2F and F2M patients, $p<0.225$.
Conclusions: In our cohort of children and adolescents with GD, ~50% were overweight or obese at their first clinic visit prior to hormonal intervention. Potential risk factors for abnormal weight gain in these patients include depression, anxiety, disordered eating and psychotropic medications. The high prevalence of overweight in transgender youth represents an important co-morbidity with the potential for adverse health consequences. How BMI is impacted by hormonal treatment in M2F and F2M pediatric patients remains to be determined.

P3-605

BICALUTAMIDE AS AN ANDROGEN BLOCKER WITH SECONDARY EFFECT OF PROMOTING FEMINIZATION IN MALE TO FEMALE (MTF) TRANSGENDER ADOLESCENTS

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Objectives: GnRH analogs are first-line treatment for halting pubertal development in gender variant youth. However, this medication is often denied by third party payors. The pure androgen receptor blocker bicalutamide represents a potential alternative approach to blocking puberty in natal males. Here, we describe the use of bicalutamide in MTF transgender patients.
Methods: Medical records for patients with gender dysphoria (GD) followed in the pediatric endocrine clinic at Riley Hospital for Children were reviewed. All MTF transgender patients treated with bicalutamide were included. Variables evaluated comprised age, duration of follow up, timing of estrogen initiation, laboratory studies and physical exam findings including change in breast Tanner stage during treatment.
Results: Of 77 patients with GD identified, 29 were MTF, of whom 14 (48%) aged 15.8 ± 1.9 years (range 12-18.4yr) were treated with bicalutamide 50 mg daily between 2013 and 2017. Of these, 3 were started on estrogen concurrently whereas 11 received bicalutamide alone, 7 of whom have returned for follow up thus far. After an average of 5.7±1.5 months, 86% of the patients (n=6) had breast development consisting of Tanner stage III in 4, Tanner stage II in 1, and Tanner stage III/II of the right and left breast in 1. The 7^{th} patient was noted to have Tanner III breasts at her 2^{nd} follow-up clinic visit 12.5 months after starting bicalutamide. LFTs were obtained on 4 patients, estradiol on 3 patients and testosterone on 2 patients while exclusively taking bicalutamide. LFTs were unremarkable and concentrations of estradiol and testosterone were 26-61 pg/mL and 524-619 ng/dL, respectively.
Conclusions: Bicalutamide is used in rare forms of precocious puberty in males and has a known side effect of gynecomastia. Here, we report the novel use of bicalutamide as a puberty blocker in MTF patients with GD in whom it also results in feminization by causing breast development. Additional studies are needed to further evaluate the potential role of bicalutamide in the therapeutic armamentarium for the treatment of transgender MTF adolescents.

P3-606

CHILDREN AND ADOLESCENTS WITH GENDER DYSPHORIA - THE ISRAELI EXPERIENCE

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Objectives: To describe patient characteristics at presentation, management and response to treatment of children and adolescents with gender dysphoria in Israel.
Methods: A retrospective chart review of 46 consecutive children and adolescents (< 18 years) with gender dysphoria referred to and followed at the Israeli multidisciplinary Pediatric Gender Dysphoria Clinic from 2013- 2017.