dominant disorder. Unilateral adrenalectomy could control the cortisol hypersecretion, however some of these patients can have subclinical Cushing Syndrome.

Pediatric Endocrinology

PEDIATRIC ENDOCRINE CASE REPORTS II

Rare X Chromosome Pericentric Inversion Associated with Ovotesticular Disorder of Sex Development.

Gil Guerra-Junior, MD, PHD[1], Ana Paula Santos, PhD[2], Flavia M. Oliveira, BSc[2], Tarsis A P Vieira, PhD[2], Nilma L V Campos, PhD[2], Marcio L. Miranda, MD, PhD[2], Mara S. Guaragna, PhD[2], Helena Fabbri-Scallet, PhD[2], Maricilda P. de Mello, PhD[2], Andrea Trevas Maciel-Guerra, MD, PhD[2].
[1]Universidade Estadual de Campinas (UNICAMP), Campinas - SP, Brazil, [2]State University of Campinas, Campinas, Brazil.

MON-056

Ovotesticular disorder of sex development (OT-DSD) is a rare condition characterized by coexistence of ovarian and testicular parenchyma, in separate gonads or in the same gonad (ovotestis), in individuals with variable degrees of genital ambiguity. Karyotype may be 46,XX (60%), 46,XY (10%), or there may be sex chromosomes abnormalities, including mosaicism, chimerism and structural anomalies (30%). A genetic origin may be identified in some subjects with normal XX or XY karyotype, but most cases are of unknown origin. Apparently balanced chromosome rearrangements (translocations, insertions and inversions) may cause truncation, deletion, inactivation or over-expression of specific genes. We report on a case of OT-DSD associated with an X chromosome inversion. **Case report:** A 3-month old girl was referred due to atypical genitalia. She was born at term to a 42 years old G3P2A1 mother and her 45 years old unrelated husband with normal weight, length and head circumference. She had normal development, no associated health problems, and family history was unremarkable. Physical examination revealed a 3.1-cm phallus with *chordee*, scrotal hypospadias, partial penoscrotal inversion and a 0.5 cm^3 right gonad palpable in the inguinal region; there were no associated dysmorphic features. At 1.5 months there were normal levels of FSH (3.09 IU/L) and LH (3.67 IU/L), and testosterone (155 ng/dL) was in the normal male range. Ultrasound revealed normal uterus and gonads were in the inguinal regions. Urethracystoscopy and vaginoscopy at 9 months revealed a urogenital sinus with high vaginal confluence. Laparoscopy and gonadal biopsies were also performed; the left gonad was an ovotestis with multiple ovarian follicles, while the right gonad was a testis. In both gonads the seminiferous tubules had only Sertoli cells. Karyotype revealed a pericentric X chromosome inversion, 46,X,inv(X)(p22.1q26)dn[20]. FISH on peripheral blood and cultured cells from the right gonad with probes for X (DXZ1) and Y (DYZ3) centromeres and *SRY* (Yp11.3 - 122 Kb) showed only two X chromosome signals. Array GH analysis (Cytoscan 750K, Affymetrix) showed a 1.3 Mb deletion distal to the short arm breakpoint (Xp22.31), which was reported as VOUS, and a 9 Mb region of LOH on chromosome 9. **Discussion:** Several cases of X pericentric inversion with different breakpoints have been reported; though phenotypes of female heterozygotes are often normal, early menopause, irregular menses, gonadal dysgenesis or sterility have been described. In this case, it is plausible that the genomic rearrangement could have affected long-range regulation of *SOX3* (located in Xq27.1) resulting in ectopic expression of this gene in the bipotential gonad. In addition, the features detected in array GH may have a role in the phenotype. Different methods to determine the exact chromosomal breakpoints and copy number variations in this region will be required.

Reproductive Endocrinology

TRANSGENDER MEDICINE AND RESEARCH

Gender Dysphoria - Data from One Hundred Patients

Maria João Ferreira, MD[1], José Luis Castedo, MD[1], Jorge Pires Pedro, MD[1], Cristina Daniela Salazar, MD[1], Cláudia Fernandes Costa, MD[2], Davide M. Carvalho, MD,PhD[3].
[1]Department of Endocrinology, Diabetes and Metabolism of Centro Hospitalar Universitário São João, Porto, Portugal, [2]IPO Porto, Matosinhos, Portugal, [3]Faculty of Medicine, University of Porto, Portugal, Sao Mamede Infesta, Portugal.

SUN-051

Introduction: Gender dysphoria (GD) is a disease in which the patient's gender identity does not correspond to their biological gender. Traditionally, sex is assigned accordingly to the sexual organs as male or female, however these patients do not identify with this classification. A biological male who identifies himself as a female is referred to as transgender female or male to female GD (MtF) and a biological female who identifies herself as a male is referred to as a transgender male or female to male GD (FtM). In our center there is a multidisciplinary consultation to approach and follow up patients with GD. Our work is an analysis of the patients who are currently attending this consultation. Methods: Retrospective study of patients attending the Sexual Medicine Group Consultation with diagnosis of GD. Results: 100 patients diagnosed with GD, of which 65% had GD FtM and 35% MtF. The median age is 25 years (± 9.5 years). Regarding the onset of symptoms, 76% of patients report that it occurred in childhood (before age 10); 20% from 10 to 18 years old and only 4% reported having occurred after 18 years. The median age for starting treatment is at 22 years (± 7.5 years). About 83% of the patients start therapy on medical advice, the remaining are self-medicated; 86% of FtM GD patients are currently on testosterone therapy; 85% of MtF GD patients are on estrogen therapy and 71% on anti-androgens. Concerning co-morbidities: 22% of patients have depression; 39% smoke; 6% are HIV-infected; 4% dyslipidemia and 2% hypertension. 47 (47%) patients underwent at least one surgical treatment; the average time to the first surgery was 2 years. Of the FtM GD patients, 23 (35%) patients underwent mastectomy, 10 (15%) hysterectomy and oophorectomy and 3 (5%) neophaloplasty; of the MtF GD patients 8 (23%) underwent augmentation mammoplasty; 7 (20%) neovaginoplasty. Two patients had a family history of GD. One case of Klinefelter's syndrome and one case of congenital adrenal hyperplasia was diagnosed during the study prior to therapy initiation.
Discussion: There was a higher prevalence of FtM compared to MtF, unlike most centers reporting a higher prevalence of MtF. Regarding the proportion of patients on medical therapy, the data are in line with data from other centers