Endocrine Abstracts

May 2025 Volume 110
ISSN 1479-6848 (online)

Joint Congress of the European Society for Paediatric Endocrinology (ESPE) and the European Society of Endocrinology (ESE) 2025: Connecting Endocrinology Across the Life Course

10–13 May 2025, Copenhagen, Denmark

Connecting Endocrinology Across the Life Course
Joint Congress of ESPE and ESE 2025
Copenhagen, Denmark. 10-13 May 2025

ESPE
European Society for Paediatric Endocrinology

European Society of Endocrinology

published by
bioscientifica

Online version available at
www.endocrine-abstracts.org

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differentiation and development of male external genitalia. Mutations in NR5A1 gene can result in a spectrum of phenotypes related to sexual differentiation (e.g., partial or complete gonadal dysgenesis, primary ovarian insufficiency, male infertility – oligo-azoospermia), reflecting its interaction with other key genes, such as GATA4, AMH, WT1, CBX2, and WNT4, affecting the regulatory network of gonadal development.
Case Report
A two-month-old infant was referred to the Endocrinology Clinic due to atypical external genitalia and micropenis. The child was born after an uneventful monitored pregnancy. Prenatal ultrasounds showed no abnormalities, and the fetus was designated as female. Delivery occurred by caesarean section at 42 weeks, with good neonatal adaptation. Due to atypical external genitalia, karyotyping revealed 46,XY and pelvic ultrasound identified testes near the scrotal sacs with no Müllerian structures. Male gender was assigned. At the two-month Endocrinology Clinic visit, the infant presented with penoscrotal hypospadias, micropenis, and testes located in the scrotum. Biochemical assessment excluded congenital adrenal hyperplasia and hormone levels were consistent with mini-puberty. The patient remained under follow-up, with serial ultrasounds consistently showing small testes with hyperechoic foci. At five years of age, molecular testing for androgen insensitivity was performed, but no mutations were identified. At 8 years, testosterone treatment (25 mg monthly for three months) was administered for micropenis, however no response was observed. Due to persistently low gonadotropin and testosterone levels, puberty induction with testosterone was initiated at 12 years, resulting in the development of pubic and axillary hair, penile growth, and the onset of acne. At 16 years, genetic testing identified a likely pathogenic variant, c.442_456del, p.(Glu148Glyfs*44), in heterozygosity, in exon 4 of the NR5A1 gene.
Conclusion
Testing for NR5A1 gene mutations should always be performed in cases of disorders of sexual differentiation, given the broad spectrum of associated phenotypes. Genetic diagnosis in these conditions enables optimised therapeutic management, clearer prognosis assessment, and genetic counselling for family members.

DOI: 10.1530/endoabs.110.EP1382

EP1383

JOINT2197

Increased alpha-glucosidase levels in sertoli cell-only syndrome: a report of three cases

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Background
Sertoli cell-only syndrome (SCOS) is one of the most common causes of non-obstructive azoospermia in infertile men. The definitive test for diagnosing SCOS is the testis biopsy. In this work, we present 3 cases of SCOS in which the alpha-glucosidase (α-GLUC) level was remarkably elevated.
Clinical Cases
Case 1: A 35-year-old male presented with primary infertility and secondary erectile dysfunction. Laboratory investigations revealed hypergonadotropic hypogonadism and total azoospermia. Testicular atrophy was confirmed on ultrasound and testicular biopsy confirmed the diagnosis of SCOS. α-GLUC levels was elevated. Genetic testing identified a partial AZFc microduplication on the Y chromosome. The karyotype was normal. **Case 2:** A 43-year-old male presented with severe osteoporosis, pathological fractures, bilateral gynecomastia, and infertility without erectile dysfunction. Azoospermia was confirmed, along with hypergonadotropic hypogonadism and bilateral testicular atrophy. Genetic screening was negative for Y-chromosome microdeletions. The karyotype was normal. A testicular biopsy revealed SCOS. α-GLUC levels was also elevated. **Case 3:** A 49-year-old male presented with primary infertility and normal erectile function. Laboratory investigations showed hyperprolactinemia without an identifiable cause, hypergonadotropic hypogonadism, and azoospermia. Testicular ultrasound confirmed bilateral testicular atrophy. A testicular biopsy confirmed the diagnosis of SCOS. The karyotype was normal. Genetic testing revealed a partial AZFc microdeletion on the Y chromosome. MRI of the hypothalamic-pituitary axis was normal. α-GLUC levels was elevated.
Discussion
SCOS also known as germ cell aplasia is characterized by azoospermia in which the seminiferous tubules of testicular biopsy are lined only with Sertoli cells. The exact diagnosis of SCOS is based on diagnostic testicular biopsy and histopathological examination. Seminal α-GLUC levels reflect the epididymal function. The diagnosis of SCOS was retained in these 3 patients thanks to the testicular biopsy. These 3 patients had an elevated α-GLUC level suggesting a possible relationship in the pathophysiology of SCOS that should be investigated by large-scale studies. Would this marker be useful in the diagnosis of SCOS in the future?.

DOI: 10.1530/endoabs.110.EP1383

EP1384

JOINT1480

Endocrine factors in the development of recurrent pregnancy loss clinical case

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Introduction
Recurrent pregnancy loss (RPL) is a polyetiological complication of pregnancy, primarily caused by dysfunction of the reproductive system. Endocrine factors play a significant role in the etiology of spontaneous miscarriages. The prevalence of endocrine factors in recurrent pregnancy loss is approximately 17%. The most common hormonal disorders contributing to RPL include ovarian hypofunction, hyperandrogenism of various origins, thyroid hypofunction, hyperprolactinemia, and thyroid pathology.
Objective
Present a clinical case with RPL.
Materials and methods
The primary documentation has been reviewed: medical history, results of clinical, laboratory, and ultrasound examinations.
Results
In January 2023, a 28-year-old woman consulted an endocrinologist to identify possible endocrine causes of RPL. Medical history revealed an irregular menstrual cycle (2537 days). She had experienced four pregnancies: the first ended in preterm delivery at 30 weeks due to placental insufficiency, while the others resulted in spontaneous miscarriages at 9, 10, and 8 weeks. Complaints included fatigue, irritability, emotional instability, headaches, and menstrual irregularities. Hormonal studies were recommended. Test results obtained the following day revealed elevated prolactin levels (63.5 ng/ml) and TSH (8 mU/L), with T4 and T3 levels within the normal range. An MRI of the brain was subsequently recommended to rule out a prolactinoma. After thorough examinations, the patient was diagnosed with idiopathic hyperprolactinemia and subclinical hypothyroidism. Dopamine agonists and hormone replacement therapy were prescribed. Four months later, the patient became pregnant, and all medications were discontinued. However, signs of a threatened miscarriage appeared at 8 weeks. Hormonal analysis revealed elevated prolactin (200 ng/ml) and TSH (6 mU/l)levels, with a decrease in inhibin A (420 pg/ml). Treatment was resumed based on these findings. The patient was closely monitored throughout the pregnancy, with periodic assessments of prolactin, TSH, and inhibin A levels. Treatment was adjusted according to hormone levels. In 2024, the pregnancy concluded with a full-term delivery at 39 weeks without complications.
Conclusion
Elevated prolactin and TSH levels are associated with decreased inhibin A, indicating an increased risk of pregnancy loss. Inhibin A may be used as a predictor of RPL development linked to endocrine factors (e.g., hyperprolactinemia and hypothyroidism).

DOI: 10.1530/endoabs.110.EP1384

EP1385

JOINT2361

The relationship between indicators of leptin and anti-Muller hormone in women suffering from infertility and obesity

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