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P32. Clinical, biochemical and genetic characterization of a 44- years old patient with primary amenorrhoea- case study of complete androgen insensitivity syndrome

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Context: Complete Androgen Insensitivity Syndrome is a congenital disorder which manifests itself by total cells resistance to androgens actions. The pathogenesis involves a defective androgen receptor gene, which is located at X chromosome (Xq11-12) and results with the development of feminine phenotype in chromosomally male individual.
Objective: The case study consists of a clinical, biochemical and genetic investigation of a patient finally diagnosed with CAIS.
Methods: Serum levels of LH, FSH, testosterone, DHEAS and estradiol were measured. Imaging tests such as pelvic ultrasound, pelvic MRI and inguinal ultrasound had been performed in differential diagnosis. Finally the chromosomal analysis of a karyotype had been done. Methode of molecular biology revealed the mutation in androgen receptor gene.
Patient: 44 years old Caucasian female was admitted to the Department of Gynecological Endocrinology in Poznan University of Medical Science because of primary amenorrhea. She had no relevant past medical or family history. Her weight was 98 kg, her hight was 164 cm and BMI was 36,5 kg/m^2. The patient had no axillary hair, Tanner stage 3 breast development, and Tanner stage 1 pubic hair growth. The external genitalia appeared normal but a short, blind vaginal pouch was detected with a depth of 3 cm.
Results: Hormonal results: Laboratory evaluation revealed FSH level 30,05 mIU/ml, LH level 24,37 mIU/ml and estradiol serum level 83,34 pg/mL. Serum testosterone level was elevated at 5,29 ng/ml and DHEAS was in normal range for the age (5,19 mmol/L).
Imaging tests: Transvaginal pelvic ultrasound revealed the absence of a uterus, fallopian tubes, and gonads.
Pelvis MRI confirmed the lack of gonads and uterus.
Finally the ultrasound of inguinal canals revealed the presence of structures with typical testicular echogenicity.
Molecular tests: Chromosomal analysis confirmed the 46XY karyotype.
Genetic tests were performed to find a specific mutation in androgen receptor gene- the substitution from cytosine to tymine in chromosome 23:66942710 position.
Conclusions: AIS is a rare disorder typically recognised in young woman during the diagnosis of primary amenorrhea. The disorder requires urgent preventive gonadectomy due to increased risk of neoplasms (seminoma being the most common). Typically hormonal replacement therapy has to be administered to

prevent osteoporosis. The psychological support for the patient and her family should be provided.

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