

## Case description

A 10 years old female patient, was referred to the endocrinology department for evaluation of short stature. A neurologist first evaluated her at 7–8 months of age following multiple consultations for mild mental retardation, developmental delays, and dysmorphic features, including a flat nose, low-set eyebrows, and small, low-set earlobes. The brain MRI at the time showed no abnormalities. Dysmorphic features and Turner syndrome-like dysembryogenesis symptoms prompted genetic evaluation. Karyotyping revealed a 46XY genotype, confirming the diagnosis of 46XY DSD. Further investigations revealed bifurcated kidneys, left kidney pyelectasis, and anomalies of the coronary vessels. Upon admission to the endocrinology department, a physical examination revealed hypertelorism of the nipples, an enlarged clitoris, a narrow vagina, and hypoplastic internal sex organs. Laboratory investigations demonstrated significantly reduced anti-Müllerian hormone (AMH) levels at 0.012 ng/ml and testosterone at 2.5 ng/dl (both below the normal range). IGF-1 was 68.2 ng/ml (IGF-1 SDS: -2.38), with a bone age of 6 years, reflecting delayed skeletal development. The hCG stimulation test results showed no increase in testosterone levels. Given the high risk of gonadal malignancy associated with 46XY DSD, diagnostic laparoscopy and gonadectomy were planned but postponed due to asymptomatic leukocyturia detected during preoperative testing. The patient now remains under close monitoring and is preparing for the surgery. Considering the presence of gonadal dysgenesis, growth hormone treatment is planned to begin after gonadectomy.

## Conclusions

This case underscores the critical importance of timely multidisciplinary evaluation in 46XY DSD patients. Early genetic testing and endocrinological referral are essential for optimizing outcomes and preventing delays in diagnosis and management. This case also highlights the importance of vigilant monitoring for gonadal malignancy, necessitating timely surgical intervention when feasible as there is a high risk of gonadal malignancy.

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## EP1343

## JOINT911

## Polycystic ovary syndrome: what we know about it?

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## Introduction and Aim

Approximately 70% of people with PCOS are undiagnosed.

The aim was to study the knowledge of PCOS among medical students.

## Materials and Methods

It was a cross-sectional study by questionnaire method. The research involved survey consisting of 15 questions, first 9 questions were about the definition, symptoms and the diagnostic criteria of PCOS; the second part was about the student's experience and opinion. The sample size was 17 students (14 females and 3 males) from international faculty of general medicine (4–5 courses) 20–25 years old with BMI between 23,5 and 32,5 kg/m<sup>2</sup>.

## Results

In “What is PCOS from your point of view?”, 58,8% answered that it is a disease, the other answered that PCOS is an ultrasound diagnosed symptom. In “What is the correct definition for PCOS?” there were significant debate about the convenient definition: 29,4% (5 students) chose that it is reproductive disorder, 35,3 (n = 6) answered it's a metabolic disorder, and the rest chose none of the above choices. In “What is the most affected age?” most of the students answered 18–38. Coming “How can you tell ovulation had happened?” 47,1% (8 students) answered that by hormonal changes and ultrasound, 29,4% (n = 5) chose by mood swings and feelings, however the rest confirmed that test is the one to know if ovulation had happened. There was similar answer in “Gallwey Score is used to test?” most of them had confirmed that they had never heard about it. Majority had answered that dysmenorrhea is the needed criteria to diagnose PCOS, only 17,7% (3 students) chose that infertility and psychosexual dysfunction is the needed one. It was a popular opinion the female should be treated from PCOS despite of her planning pregnancy, so 64,7% (11 students) chose that any female who has dysmenorrhea should be suspected with PCOS. Question 10 was only for females, if they use any application to check their menstrual cycle, however in answering question 4 about the normal cycle length all had the same answer 21–35. Most the students confirmed that the female should check her hormone levels every 3 months. Apparently, most of them think that PCOD (disorder) is different from PCOS (syndrome). There was difference in opinions about who should treat PCOS: 58,8% (n = 10) chose gynecologist, 23,5% (n = 4) added their own opinion and the others chose the endocrinologist.

## Conclusion

The most of the female students use menstrual calendar, however they didn't know what is the normal cycle length. Although every one of the medical students had different point of view and thoughts about the best treatment for PCOS, but all had confirmed that PCOS is a huge misunderstanding topic.

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## EP1344

## JOINT2104

## Diabetic turner syndrome patients – conclusions from the bulgarian diabetes registry

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## Background

The Bulgarian healthcare system requires individual compulsory health insurance administered by the National Health Insurance Fund (NHIF), operating with the health data of 2.75 million Bulgarian women. The Bulgarian Diabetes Register (BDR) collected all pseudonymized NHIF electronic health records of patients with diabetes mellitus type 1 or type 2 (T1DM or T2DM), with the latest dataset being from 2018 (1). BDR includes outpatient NHIF records from all primary care providers and specialists in secondary care nationwide for every visit of a patient suffering from DM.

## Methods

The available data about diabetic patients with Turner syndrome (TS) (ICD-10 Q96, ORPHA:881) from the BDR were extracted and analyzed. The collected information included age, type of diabetes, diabetes complications, treatment, concomitant diseases, and a number of medical consultations yearly.

## Results

A total of 19 diabetic patients (median 40 [13–69] years) with TS have been found in the BDR database. 84.2% of patients were with DM2, while 15.8% were with DM1. Diabetic neuropathy was found in 7 (36.8%) of the patients. The TS patients showed significant co-morbidity and a high number of consultations with general practitioners and/or clinical specialists yearly - an average of 14 (2–29) in 2018.

## Conclusions

The lower-than-expected prevalence of diabetic TS patients in the BDR might result from TS or DM underdiagnosis. Diabetic TS patients have a high prevalence of complications and co-morbidities and require high healthcare resources. More efforts should be made by the public health system in the country to ensure the proper diagnosis of TS. Furthermore, regular estimation of carbohydrate metabolism in TS patients is paramount for the early diagnosis of DM in the affected women and the prevention of complications and high medical costs.

## References

1. Krastev E, Markov E, Abanos S, Krasteva R, Tcharaktchiev D. Mapping the Bulgarian Diabetes Register to OMOP CDM: Application Results. *Stud Health Technol Inform.* 2024;313:28–33.

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## EP1345

## JOINT1135

## Etiologic distribution of adult-onset male hypogonadism in the modern era: retrospective cohort study from finnish tertiary center

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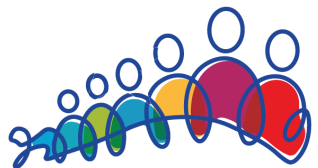
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