be brought about by various factors such as urine volume, medication interference and certain foods.

CASE

We report a case of a 67-year-old male with Hepatitis B and smear-positive pulmonary tuberculosis on maintenance treatment with rifampicin and isoniazid, who was referred for left adrenal incidentaloma from CT of the hepatobiliary system. He denied any paroxysmal symptomatology of phaeochromocytoma and was normotensive. 24-hour urinary metanephrines revealed significantly elevated normetanephrine (14 times the upper limit of normal [30.15 µmol/day]), with normal metanephrine and 3-methoxytyramine levels. The adrenal CT demonstrated a left adrenal mass measuring 2.7 x 1.4 x 2.6 cm, 32 Hounsfield units (HU), with absolute and relative washout of 62.8% and 19.6%, respectively, indicating an indeterminate adrenal mass. The patient was diagnosed with left phaeochromocytoma and underwent laparoscopic left adrenalectomy with Phenoxybenzamine cover. However, the histopathological findings revealed multiple granuloma formation, with special stains negative for acid-fast bacilli, suggestive of chronic changes of right adrenal tuberculosis (non-active) and no features of phaeochromocytoma. Thoracic and abdominopelvic CT scans showed no evidence to suggest paraganglioma, which might contribute to elevated normetanephrine levels. A post-operative repeat 24-hour urine metanephrine came back normal. This repeated sample was taken after the patient completed tuberculosis treatment (including rifampicin). Some reports recognised rifampicin interference with urinary metanephrine measurement as it is eluted with normetanephrine, causing significantly elevated levels. These findings correlate with this patient as urine normetanephrine returned to normal once he was off rifampicin.

CONCLUSION

Histopathological findings of the left adrenal mass were suggestive of post-adrenal tuberculosis rather than phaeochromocytoma. Rifampicin was found to be an interferent in urine metanephrines measurement, which led to falsely elevated normetanephrine levels with no catecholaminergic signs or symptoms.

EP_A087

A RARE CASE OF TURNER MIMICKER

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INTRODUCTION/BACKGROUND

Primary amenorrhea and delayed puberty are frequently encountered in primary care, prompting suspicion of Turner syndrome, especially in cases with short stature. This case underscores the importance of considering Swyer syndrome even when significant growth impairment is present.

CASE

A 28-year-old phenotypic female, born of a non-consanguineous union, presented with primary amenorrhea and a short stature of 1.31 meters. Physical examination revealed absent secondary sexual characteristics (Tanner stage 1). External genitalia were unambiguously female. Bone age assessment identified significant delay, corresponding to a 15-year-old. The hormonal evaluation showed hypergonadotropic hypogonadism. Thyroid function and insulin growth factor-1 levels were normal. Pelvic MRI demonstrated an atrophic uterus, absent fallopian tubes and ovaries. Karyotype analysis confirmed a 46, XY genotype, consistent with Swyer syndrome. Following pubertal induction for 3 months, she developed regular menstruation and progression to Tanner stage 3.

Swyer syndrome is a rare disorder of sex development featuring female phenotype, hypergonadotropic hypogonadism and streak gonads. While 15-20% of cases result from SRY gene mutations impairing testis-determining factor function, other genes have also been implicated. Swyer syndrome classically causes tall stature from estrogen-deficient delayed epiphyseal fusion. However, our case exhibited profound short stature and severely delayed bone age, explained by the complete prepubertal estrogen deprivation abolishing both growth spurt and fusion. Additional factors, like SHOX gene variations, may have contributed to her growth impairment. Diagnostic complexity arose from initial Turner syndrome overlap; however, the absence of other Turner stigmata and 46, XY karyotype confirmed Swyer syndrome. This emphasises karyotyping's diagnostic importance in primary amenorrhea with hypergonadotropic hypogonadism, regardless of phenotype. Hormone replacement therapy remains crucial for puberty induction, bone health and cardiovascular protection.

Adult E-Poster

CONCLUSION
Swyer syndrome may mimic Turner syndrome in cases of primary amenorrhea with short stature. Accurate diagnosis requires comprehensive hormonal, imaging and genetic evaluation beyond clinical phenotype alone.

EP_A088

UNRAVELLING THE MYSTERY: A CASE OF ATYPICAL DIABETES WITH HEPATIC AND RENAL CLUES TO HNF1B DEFICIENCY

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INTRODUCTION/BACKGROUND
Hepatocyte nuclear factor 1β (HNF1B) deficiency associated with MODY-5 is increasingly recognised as a multifaceted syndrome with diverse manifestations. We present a suspected case initially misdiagnosed as type 1 diabetes with autoimmune hepatitis.

CASE
A 14-year-old male with learning disability was admitted for insulin initiation when he presented with osmotic symptoms with hyperglycaemia and ketonuria. He reported no family history of diabetes. HbA1c was 18.5% and LFTs were deranged (AST 74, ALT 209 and ALP 451 IU/L). He has some dysmorphic facial features. Despite good glycemic control on intensive insulin therapy, his liver enzymes remained elevated (8-17x ULN) with normal ferritin, ceruloplasmin and viral panel. Abdominal ultrasound showed normal liver and spleen but detected bilateral medullary nephrocalcinosis. The liver biopsy showed mild periportal hepatitis. He was treated for autoimmune hepatitis with prednisolone and azathioprine. Subsequent investigations revealed negative diabetes (anti-GAD, ICA, IA2), hepatic (ANA, smooth muscle, LC1, LKM and mitochondrial) autoantibodies and normal serum immunoglobulins. The absence of diabetes-related autoantibodies, coupled with multisystem involvement (pancreas, liver, kidney, neurocognitive and dysmorphism), raised the suspicion of HNF1B mutation. Although genetic confirmation was not feasible, further investigation with elevated C peptide (1652 pmol/L) and persistent hypomagnesemia (0.4 to 0.55 mmol/L) further substantiated this hypothesis. Immunotherapy was withheld. He remained well with fluctuating liver function on follow-up 5 years since the initial presentation.

CONCLUSION
This case underscores the diagnostic complexity of HNF1B deficiency, a rare monogenic diabetes subtype accounting for ~6% of MODY. Despite an autosomal dominant inheritance pattern, de-novo mutation accounts for 50% of cases. Lack of family history does not preclude the diagnosis. Diagnostic clues include multisystem involvement, which is rarely found in other MODY subtypes. Hypomagnesemia is another common feature. Early recognition is essential for individualised management, avoidance of mismanagement, monitoring for other organ involvement or complications and genetic counselling.

EP_A089

TWIN-TWIN TRANSFUSION SYNDROME ASSOCIATED MATERNAL HYPERTHYROIDISM

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INTRODUCTION/BACKGROUND
Pregnancies complicated by twin-twin transfusion syndrome (TTTS) are associated with elevated human chorionic gonadotropin (hCG) compared to uncomplicated twin pregnancies. Studies have shown a positive correlation between hCG and free thyroxine (FT4) in TTTS, thereby increasing the risk of maternal hyperthyroidism. This case report describes a twin pregnancy complicated by TTTS, where maternal hyperthyroidism developed prior to fetoscopic laser ablation (FLA).

CASE
We present a 36-year-old female with a twin pregnancy complicated by TTTS. She was diagnosed with gestational transient thyrotoxicosis (GTT) at 10 weeks of gestation with thyroid stimulating hormone (TSH) of 0.01 mIU/L, FT4 of 24.8 pmol/L and triiodothyronine (T3) of 3.8pmol/L. She had negative thyroid-stimulating hormone receptor antibodies and a normal neck ultrasound. Clinically, she has no goitre or thyroid eye disease. At 15 weeks of gestation, her FT4 decreased to 14.3 pmol/L while TSH remained suppressed. She did not receive any anti-thyroid drugs (ATDs) during the first trimester. She was admitted at 22 weeks of age of gestation for FLA due to TTTS stage 1. Upon admission, she complained of palpitations, and the cardiac monitor showed sinus tachycardia with a heart rate of 123 bpm. Her TSH was <0.008 mIU/L, FT4 was increased to 21 pmol/L and hCG of >225,000U/L. Due to hyperthyroid symptoms, she was treated with carbimazole and beta-blocker prior to FLA. Her carbimazole dose was reduced at 25 weeks of gestation as FT4 dropped to 13.2 pmol/L. It was then