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.

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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

discontinued at 28 weeks of gestation (FT4 11.25 pmol/L; TSH 0.11 mIU/L). She underwent emergency hysterectomy at 28 weeks of gestation due to TTTS progression to stage 4.

CONCLUSION

GTT in twin pregnancies typically resolve by the end of the first trimester. A sustained FT4 increase should raise suspicion for TTTS. ATDs should be considered due to the risk of TTTS-associated maternal hyperthyroidism, as it may persist until successful FLA.

EP_A090

WHEN LIGHTNING STRIKES TWICE: A CASE OF METACHRONOUS INVASIVE BREAST CARCINOMA AND PAPILLARY THYROID CARCINOMA IN A FEMALE FILIPINO PATIENT

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INTRODUCTION/BACKGROUND

Triple-negative invasive ductal carcinoma is a more aggressive type of breast cancer that poses therapeutic challenges. Papillary thyroid carcinoma is generally indolent but has shown aggressive behaviour among Filipinos. As primary carcinomas, both tend to have a good prognosis with early detection and management. However, failure to anticipate a secondary malignancy, when one occurs after the other, can turn a treatable journey into a devastating outcome.

CASE

A 45-year-old female presented with a movable left breast lump 3 years ago. Biopsy confirmed invasive ductal carcinoma with a negative ER/PR/HER2 on immunohistochemistry. She eventually underwent a modified radical mastectomy of the left breast and staged as 2B (T2N1M0) due to the absence of lympho-vascular space invasion and distant metastasis. She completed eight cycles of adjuvant chemotherapy with Doxorubicin and Cyclophosphamide. Post-chemotherapy surveillance confirmed the absence of metastasis. A 2 x 2 cm thyroid nodule was detected on the left anterior neck two years later during routine follow-up. Ultrasound revealed a lobulated solid hypoechoic wider-than-tall nodule in the superior pole of the left lobe (TI-RADS 5). The patient was clinically and biochemically euthyroid. Ultrasound-guided fine needle biopsy identified the presence of Papillary Thyroid Carcinoma (Bethesda Category VI). As such, the patient underwent a total thyroidectomy. Final histopathologic studies confirmed a classic subtype of Papillary Thyroid Carcinoma (ATA Low Risk) without lymphatic, perineural, extrathyroidal invasion and regional lymph node metastasis. Post-operative high-dose radioactive iodine was administered to eliminate any residual thyroid tissue. She was then maintained on levothyroxine suppression and continuously monitored for tumour recurrence.

CONCLUSION

As better understanding of tumorigenesis has revolutionised cancer screening and management, the metachronous coexistence of breast and thyroid carcinoma highlights the importance of multidisciplinary care and vigilant screening for secondary malignancies. Overexpression of estrogen and progesterone, together with shared environmental and genetic factors in breast cancer, have been shown to promote thyroid tumorigenesis and progression.

EP_A091

EXPERIENCE OF CINACALCET TREATMENT DURING PREGNANCY IN PRIMARY HYPERPARATHYROIDISM

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INTRODUCTION/BACKGROUND

Primary hyperparathyroidism (PHPT) in pregnancy is rare but associated with high maternal (68.6%) and foetal (80%) complications, including pre-eclampsia, miscarriage and intrauterine growth restriction. The risks are directly related to the severity of the disease and the serum calcium level. We describe two cases with differing clinical outcomes based on the timing of diagnosis and intervention.

CASE

A 30-year-old gravida 3, para 2, presented with maternal tachycardia at 27 weeks of gestation. On work-up, the patient was incidentally found to have hypercalcaemia. ECG showed a shortened QTc. Biochemically, her calcium was 2.99 mmol/L (Reference Value [RV]:2.2-2.7 mmol/L), phosphate 0.7 mmol/L (RV: 0.8-1.45 mmol/L) and intact PTH level of 12.3pmol/L (RV: 1.58-6.03), suggestive of parathyroid (PTH) dependent hypercalcaemia. Ultrasound showed an enlarged right parathyroid gland. Despite IV hydration, hypercalcaemia persisted, leading to cinacalcet initiation at 29 weeks. At 30 weeks, calcium was highest at 3.05 mmol/L. She was treated with subcutaneous salmon calcitonin (5 mg/kg/dose), which was given twice daily, and cinacalcet was titrated up to 75 mg/day. Her calcium