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WHEN INFERTILITY UNVEILS UNIFYING DIAGNOSIS: AN INSIGHT TO THE DISEASE

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INTRODUCTION/BACKGROUND
Infertility involves investigation for primary or secondary causes. The presence of hypergonadotropic hypogonadism signifies premature ovarian failure (POF). When POF is accompanied by other endocrinopathies, a suspicion of underlying autoimmunity is possible. Autoimmune Polyglandular Syndrome (APS) is one of the rarer polyendocrinopathies that have multiple subtypes and clinical manifestations, including POF. We present a patient with a five-year history of diabetes and Graves' disease (in remission) who came to the fertility clinic due to secondary amenorrhea.

CASE
A 36-year-old female with a background history of diabetes and Graves' disease since 2018 presented to the gynaecology team in 2021 with primary infertility. Her diabetes was diagnosed as she presented with osmotic symptoms and was then treated accordingly with a combination of insulin and oral glucose-lowering medicine by a primary care physician. The diagnosis of Graves' disease was made by the presence of typical thyrotoxic symptoms and a positive Anti-Thyroid Peroxidase Antibody (TPO). She was prescribed carbimazole and achieved remission after one year. With regard to primary infertility, she experienced oligomenorrhea, which then progressed to amenorrhea within two years. Her serial Follicle-Stimulating Hormone (FSH) and Luteinizing Hormone (LH) levels were noted to be in the menopausal range, while her progesterone was indicative of anovulation. She was then prepared for in-vitro fertilization (IVF). Suspicion of APS arose when she was admitted for diabetic ketoacidosis. The diabetes autoantibody panel was significantly positive – Anti-Islet Cells (ICA) and Anti-Glutamic Acid Decarboxylase (GAD); hence, a diagnosis of APS was made.

CONCLUSION
Manifestations of APS vary and, most of the time, will present multiple endocrine dysfunctions within a period of time. Diagnosis and management are clinically challenging. Therefore, it may be practical to screen for other autoimmune disorders in such patients periodically.

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THE LIVER VERSUS BONE CONUNDRUM IN ISOLATED RAISED ALKALINE PHOSPHATASE LEVEL

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INTRODUCTION/BACKGROUND
Alkaline Phosphatase (ALP) is an enzyme that catalyses the hydrolysis of organic phosphate esters. It is concentrated in the liver, bone, kidney, intestinal mucosa and placenta. When ALP is elevated in isolation, isoenzyme studies using electrophoresis can confirm the source. Here, we describe a case of a patient with an incidental finding of elevated ALP.

CASE
A 30-year-old female with no known medical illness was referred to the endocrine clinic for an incidental finding of persistently elevated ALP detected during routine blood investigation. She did not report any fractures or myopathy. There were no symptoms or risk factors for liver or connective tissue disease.

Clinical examination was unremarkable, with no blue sclera, bony deformities or stigmata of chronic liver disease. Her growth was normal, achieving her mid-parental height of 150 cm. Her laboratory results showed isolated elevation of ALP 226 U/L (N: 45-129) with normal gamma-glutamyl transferase 11 U/L (N: <38), alanine transaminase 11 U/L (N: 10-49), aspartate transaminase 22 U/L (N: <34), phosphate 1.24 mmol/L (N: 0.78-1.65), calcium 2.4 mmol/L (N: 2.2-2.6), haemoglobin 136 g/L (N: 120-150), white blood cell 9.4 10^9/L (N: 4-10) and platelet 230 10^9/L (N: 150-400). Liver ultrasonography did not reveal any abnormalities.

Further investigation showed a mildly raised parathyroid hormone of 9.4 pmol/L (N: 1.96-8.49) with a low 25-OH Vitamin D level at 31 nmol/L (N: >50). She was started on cholecalciferol. Parathyroid hormone and Vitamin D became normal after treatment with cholecalciferol. Her bone scan and bone density were normal. We excluded Macro-ALP by performing a polyethylene glycol precipitation test. Subsequently, ALP Isoenzyme electrophoresis was done, confirming predominant ALP from the liver at 73% (N: 18-

85%), 14% from bone (N: 14-68%), 13% from the intestine (N: <18%). As the patient was asymptomatic and had normal liver function, the hepatologist decided to monitor her biochemically and clinically.

CONCLUSION
Patients with elevated ALP should be thoroughly investigated and examined to rule out the common treatable causes. In cases of isolated raised ALP, isoenzyme electrophoresis could identify the source with the highest accuracy.

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LIPOPROTEIN CHAOS

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INTRODUCTION/BACKGROUND
Hypertriglyceridemia pertains to blood triglyceride values greater than 2.0 mmol/L. Familial combined hyperlipidaemia, residual dyslipidaemia in well-controlled Type 2 DM and familial hypoalphalipoproteinemia are common hereditary disorders associated with hypertriglyceridemia.

CASE A
A 30-year-old female with Type 2 DM, admitted for uncontrolled DM and hypertriglyceridemia, with the following laboratory results: TG of 82.9 mmol/L (0.0-1.7 mmol/L). T Cholesterol (TC) of 14.97 mmol/L (0.0-5.2 mmol/L), non-HDL 14.48 mmol/L, LDL unmeasurable (0.0-1.95 mmol/L). She was initiated on insulin infusion, with target glucose of 8-10 mmol/L, medium chain TG (MCT) Oil 5 mls TDS, Rosuvastatin 20 mg ON, T. Fenofibrate 145 mg OD, Omega-3 1500 mg TDS. TG level decreased from 82 mmol/L to 2.7 mmol/L within one week and remained low during follow-up.

CASE B
A 48-year-old male with no comorbidities presented with left-sided weakness and facial asymmetry. He was treated as a case of cerebrovascular accident. He was incidentally noted to have hypertriglyceridemia of 11.5 mmol/L. TCl and LDL of 17.6 mmol/L and 11.7 mmol/L, respectively. Insulin infusion was initiated with fenofibrate 145 mg OD, atorvastatin 80 mg ON and Omega 3 capsules- 1 g 3 times daily. Upon discharge, his TC level was 6.3 mmol/L with TG of 4.8 mmol/L and LDL of 3.4 mmol/L. TG levels were 3.75 mmol/L during his follow-up visit with the same treatment.

CASE C
A 30-year-old female was diagnosed with hypertriglyceridemia and type 2 diabetes mellitus in the young. On admission, TC was 8.63 mmol/L, TG >12.4 mmol/L, LDL (lipaemic sample), non-HDL 8.11 mmol/L. She was initiated with insulin infusion, T. Fenofibrate 145 mg OD and Omega-3 FFA 2 g TDS. She was co-managed with a dietitian for a low TG diet with the addition of MCT oil of 8 ml BD, reduced to 5 ml BD due to intolerance. TG levels remained >12.4 mmol. After 48 hours, oral niacin 500 mg OD was added. During follow-up, TG level reduced to 3.64 mmol/L, with TC of 3.54 mmol/L and LDL of 0.82 mmol/L.

CONCLUSION
Management of hypertriglyceridemia is somewhat debatable, with some familial cases requiring plasma exchange. However, in the 3 case reports presented above, management was successful with insulin infusion, omega 3 FFA, MCT oil and statins.

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MEN2A

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INTRODUCTION/BACKGROUND
Multiple endocrine neoplasia (MEN) 2A is a rare inherited syndrome with manifestations depending on the specific RET mutation. Classical MEN2A is characterised by medullary thyroid cancer (MTC), pheochromocytoma and primary parathyroid hyperplasia.

CASE
We report a case of a 43-year-old female whose initial presentation was left flank pain, with an ultrasound showing hydronephrosis. CT scan showed a bilateral adrenal mass (>4 cm) with central necrosis. Her serum CEA and 24-hour urine metanephrine were markedly elevated. She did not present with the classical triad of pheochromocytoma. She then underwent bilateral adrenalectomy with histopathologic examination confirming pheochromocytoma. Postoperatively, she was started on fludrocortisone and hydrocortisone. A neck ultrasound was done, revealing a TR5 thyroid nodule. FNAC was done, and results showed medullary thyroid carcinoma. She was scheduled for a total thyroidectomy.

CONCLUSION
Patients with MEN might present with atypical symptoms with no positive family history. The diagnosis of pheochromocytoma will lead the clinician to investigate further to rule out MEN2A. Although MTC is usually the first manifestation of MEN2A, our patient did not present with related symptoms. Definitive treatment