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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°/L (N: 4-10) and platelet 230 10°/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-