

# 14<sup>th</sup> World Congress on Endocrinology & Diabetes

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## Hyposplenism and autoimmune hepatitis: Novel associations of Autoimmune Polyendocrinopathy Candidiasis and Ectodermal Dystrophy (APECED)

**Background:** Autoimmune polyendocrinopathy candidiasis ectodermal dystrophy (APECED) is a rare monogenic autosomal recessive syndrome caused by mutation in the AIRE gene located on chromosome 21q22.3. The phenotypic expression is variable but classically includes chronic mucocutaneous candidiasis, chronic hypoparathyroidism and hypoadrenalism. Studies demonstrate a prevalence of autoimmune hepatitis (AIH) ranging from 4 – 12%, and prevalence of hyposplenism of approximately 16%. 1,2

**Clinical Case:** This 29 year old gentleman was diagnosed with AIH at 6 years. This was followed with multiple presentations secondary to symptomatic hypocalcaemia resulting in a diagnosis of autoimmune hypoparathyroidism at 11 years. At 12 years he developed primary adrenal insufficiency and subsequent genetic analysis revealed he was homozygous for a deletion in the AIRE gene, consistent with diagnosis of APECED. Subsequent genetic screening confirmed AIRE mutations in a relative, and enquiry revealed his sister has suggestive clinical features.

At his initial clinic review in adult endocrine clinic at 19 years his diagnoses included the classic triad of hypo-parathyroidism, primary adrenal insufficiency, mucocutaneous candidiasis with AIH and nail dystrophy. At this time he was on a regimen of One alpha-calcitriol, hydrocortisone, fludrocortisone, azathioprine and Mycostatin. From 2006 – 2011 he was admitted on a number of occasions with progressive liver disease, resulting in referral for liver transplant. He had regular dental review due to the risk of oral mucocutaneous malignancies associated with APECED. In late 2011 during an admission with decompensation of liver disease it was noted he had an elevated LDH 384 I.U. /l (120 – 220) and bilirubin (total) 121 mol/l (5-24), a blood film was ordered for evaluation of autoimmune haemolytic anaemia which demonstrated Howell Jolly bodies, target cells, polychromasia, target cells and anisopoikilocytosis consistent with hyposplenism. On review of multiple previous abdominal USS his spleen was consistently small measuring 6cm in 2011 (12 – 15cm), and difficult to visualise in 2008. He was diagnosed with functional hyposplenism and treated as per guidelines with pneumococcus, haemophilus, meningitis C, influenza vaccinations and penicillin prophylaxis after a period of time.

**Discussion:** This case highlights the clinical heterogeneity associated with APECED and the importance of knowing its many associations in order to avoid missing life threatening diagnoses.<sup>3</sup> In this case there was a 12 year gap from diagnosis of APECED to recognition of his asplenia and thus initiation of an appropriate vaccination schedule.

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