In the subsequent years, both developed new symptoms of recurrent vomiting and dysphagia, suggesting achalasia. Subsequently, patient 2 had barium swallow study done which confirmed an esophageal dysmotility disorder. Retrospectively, both patients were also found to have absent tear production from a very young age, signifying alacrima.

Patient 1 managed to undergo Whole Exome Sequencing (WES) study and a homozygous variant of uncertain significance (VUS) was identified in the AAAS gene (cDNA: NM_015665.6:c.1087G>A). In view this is the gene of interest that fits into the clinical picture, the mutation was likely pathological.

CONCLUSION

High index of suspicion is required to diagnose this rare entity. Unexplained cases of adrenal insufficiency should be carefully evaluated for signs and symptoms of alacrima and achalasia.

EP_P009

AUTOSOMAL DOMINANT, NON-AUTOIMMUNE, CONGENITAL HYPERTHYROIDISM

https://doi.org/10.15605/jafes.038.S2.138

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INTRODUCTION/BACKGROUND

Primary non-autoimmune hyperthyroidism is a rare cause of neonatal hyperthyroidism. This results from an activating mutation in the thyrotropin-receptor (TSHR). TSHR is a G-protein coupled receptor and is found primarily in the thyroid gland. It is also present in adipocytes, fibroblasts and bone cells. It can be inherited in an autosomal dominant manner or occur sporadically as a de novo mutation. Affected individuals display a wide phenotype from severe neonatal to mild subclinical hyperthyroidism. The severity of the hyperthyroid symptoms is variable and phenotype differences have been described in subjects harbouring the same mutation.

CASE

A 10-month-old female was born late preterm at 36 weeks via spontaneous vertex delivery with birth weight of 2.5 kg. Antenatally, the mother was asymptomatic for hyperthyroidism. There was no family history of thyroid or autoimmune diseases. The baby was referred at birth with deranged cord TSH of 0.05 miu/L. Repeat thyroid function tests at day 7 of life demonstrated an elevated T4 of 27.6 pmol/L and low TSH of 0.01 miu/L. She had persistently high free T4, hence, was started on carbimazole at 3 months old. She was also asymptomatic for hyperthyroidism. Her growth parameters were appropriate for age. She does not have dysmorphism. She had normal heart rate, blood pressure, and tone. She did not have a goiter.

Her thyrotropin receptor autoantibodies were negative. Her neck ultrasound revealed a normal thyroid gland with normal vascularity, with no focal thyroid lesion seen. Genetic test revealed a heterozygous pathogenic variant in TSHR NM_000369.5:c.1891T>G (NP_000360.2:p.Phe631Val). During the course of follow-up, she was well and remained asymptomatic.

CONCLUSION

Although rare, TSHR gene mutation should be considered in an infant who presents with hyperthyroidism with a negative autoimmune screen and a negative maternal history of thyroid disorders. Early diagnosis is important so as to rapidly initiate anti-thyroid therapy and manage the thyrotoxicosis, to potentially avoid an enlarging goiter, and to prevent neurocognitive delays in young children with nonautoimmune hyperthyroidism.

EP_P010

DIABETIC KETOACIDOSIS WITH MELIOIDOSIS: A CASE REPORT

https://doi.org/10.15605/jafes.038.S2.139

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INTRODUCTION/BACKGROUND

Paediatric melioidosis is uncommon yet is associated with high morbidity and mortality in severe disease particularly in immunocompromised patients. Reports of melioidosis in paediatric diabetes are scarce. We present two patients with melioidosis who presented with diabetic ketoacidosis (DKA) in our centre.

CASE

Patient A is a 15 year-9-month-old female with underlying type 1 diabetes (T1D). She presented with mild DKA (serum glucose: 25 mmol/L, serum ketone: 3.9 mmol/L venous pH: 7.30, HCO_3- : 13 mmol/L) associated with fever and symptoms of upper respiratory tract infection (URTI) for

4 days. She remained febrile despite 2 courses of intravenous (IV) amoxicillin-clavulanic acid and oral erythromycin. A chest radiograph at day 9 of illness showed collapsed consolidation of the left upper lobe of the lung.

She was noted to have hepatosplenomegaly on physical examination at day 12. Abdominal ultrasound revealed multiple well-defined splenic microabscesses. Routine blood and respiratory cultures were negative, but serum IgM titres for melioidosis were positive. She was treated with IV meropenem and oral trimethoprim-sulfamethoxazole.

Patient B is a 12-year-old female who presented with prolonged fever and newly diagnosed DKA (serum glucose: 30.3 mmol/L, serum ketone: 3.5 mmol/L venous pH: 7.24, HCO_3- : 12.6 mmol/L). Her fever persisted despite DKA resolution. She developed septic shock needing intensive care admission due to severe pneumonia with bilateral pleural effusion. CT thorax and abdomen showed consolidated changes in the lungs and multiple abscesses in the liver and spleen. Blood cultures grew *Burkholderia pseudomallei* which confirmed melioidosis infection. She was treated with 6 weeks of IV ceftazidime, meropenem and trimethoprim-sulfamethoxazole.

CONCLUSION

Prolonged fever in children presenting with DKA must be thoroughly investigated. Melioidosis is uncommon, however, it needs to be ruled out to ensure adequate treatment of patients with immunocompromised status.

EP_P011

HASHIMOTO'S THYROIDITIS WITH SYSTEMIC INVOLVEMENT: A CASE REPORT

https://doi.org/10.15605/jafes.038.S2.140

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INTRODUCTION/BACKGROUND

Hashimoto's thyroiditis is the most common cause of acquired primary hypothyroidism in children. It is an autoimmune disease involving cell and antibody-mediated immune processes, leading to progressive fibrosis. Severe hypothyroidism may have variable clinical manifestation mimicking other multiorgan dysfunction.

CASE

We report an 8-year-old male who presented to our centre with a 4-month history of intermittent facial and lower limb swelling associated with unintentional weight gain, cold intolerance, easy fatigability, and regression in school performance. There was no family history of thyroid disease or autoimmune disorder. He visited the healthcare clinic 2 weeks prior to presentation for an upper respiratory tract infection however hypothyroid symptoms were not addressed. He was a short male with weight of 29.7 kg (75^{th} centile) and height of 119 cm (10^{th} centile) with evidence of faltering growth and coarse facies. He has a diffuse goitre measuring 8 x 3 cm (length x width) associated with thyroid acropachy, bilateral pretibial myxoedema and bradyarrhythmia (mean heart rate 56/min). His biochemical results showed a markedly elevated TSH 2233 mIU/L with FT4 2pmol/L, anti TPO >1000 IU/ml and anti-TG antibody 53.7 IU/ml. He was started with oral levothyroxine 25 mcg daily (0.8 mcg/kg/day) and the dose was titrated up slowly to 50 mcg daily (1.7 mcg/kg/day) over 4 weeks. He received one stress dose of intravenous hydrocortisone 100 mg (100 mg/m^2/dose) on the day of admission due to hypotension upon starting thyroxine. His status of adrenal insufficiency has not been ruled out. There were no other complications of myxoedema coma.

CONCLUSION

We report a case of missed hypothyroidism despite frank symptoms and signs. This was the highest TSH reported in our centre and likely in Malaysia. Prolonged untreated Hashimoto's thyroiditis is associated with high morbidity and mortality risk. Initiating treatment must be done cautiously to prevent crisis and complications.