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

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Vivian Lai How Mun, Janet Hong Yeoh Hua, Nalini M. Selveindran
Hospital Putrajaya, Putrajaya, Malaysia

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

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Nur Syafiqah Hamizi,[1,2] Jia Jian Tang,[1,2] Qiao Yun Lee,[1] Mia Tuang Koh,[1] Anna Marie Nathan,[1,2] Siti Zarina Yaakop,[1] Muhammad Yazid Jalaludin,[1,2] Azriyanti Anuar Zaini,[1,2] Nurshadia Samingan[1,2]
[1]Department of Paediatrics, University Malaya Medical Centre, Kuala Lumpur, Malaysia
[2]Department of Paediatrics, Faculty of Medicine, Universiti Malaya, Kuala Lumpur, Malaysia

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