Paediatrics E-Poster Presentation

EP_P001

THYROID HORMONE RESISTANCE SYNDROME (THR): A CASE REPORT

https://doi.org/10.15605/jafes.038.S2.130

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INTRODUCTION/BACKGROUND
Thyroid hormone resistance syndrome (THR) is a rare condition caused by defects in either one of the two thyroid hormone receptors which leads to tissue unresponsiveness to circulating thyroid hormones. The presentations vary depending on which receptor is affected.

CASE
A 3-year-old female who had a couple of admissions for tonsillitis was incidentally found to have failure to thrive, global developmental delay, intermittent tachycardia, and family history of goitre. She had soft dysmorphism, a baseline heart rate of 80 per min, no obvious goitre, and no skeletal dysplasia. Systemic examinations were unremarkable. Serial thyroid function tests (TFT) showed persistently high thyroid stimulating hormone (TSH) and FT4. TFT samples were sent to different biochemical laboratories and the results were similar. Autoantibody screening such as thyroid receptor antibodies, antithyroglobulin antibodies, and thyroid peroxidase antibodies were all negative. Her liver function, creatine kinase and lipid profile were normal. Thyroid ultrasound showed homogeneous enlargement of both thyroid lobes with increased vascularity within. MRI of the brain and the pituitary gland was normal which ruled out a TSHoma. Both her and her father have mutation R243W in the thyroid hormone beta gene thus confirming the diagnosis of THR-beta.

CONCLUSION
Diagnosis of THR was challenging in view of its rarity, wide spectrum of presentations, and lack of awareness among physicians.

EP_P002

A RARE CASE OF NEONATAL DIABETES WITH INSULIN GENE (INS) MUTATION: A CASE REPORT

https://doi.org/10.15605/jafes.038.S2.131

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INTRODUCTION/BACKGROUND
Neonatal Diabetes (NDM) is a rare condition that affects 1 in every 100,000-500,000 livebirths. Certain individuals with genetic mutations in NDM can be managed with sulfonylurea therapy while for others, insulin remains the mainstay of treatment. We report an infant having NDM with insulin gene (INS) mutation.

CASE
We report a small for gestational age (SGA) female born at 39 weeks via spontaneous vaginal delivery (SVD) with birth weight of 2 kg. Her parents are non-consanguineous.

She presented with fever, respiratory distress, and vomiting at 3 weeks old. She was dehydrated with 11% weight-loss. Blood investigations revealed metabolic acidosis with pH of 6.92, serum bicarbonate of 5.7 with high anion gap of 25 mmol/L, hyperglycaemia with blood sugar level of 66.15 mmol/L, and blood ketone of 2.7 mmol/L. She had concurrent inappropriately low c-peptide of 54 pmol/L (367-1467) and negative insulin autoantibodies. The genetic result showed heterozygous (p.Pro9Arg) variant in INS gene.

She was treated with intravenous fluid therapy and insulin infusion. After 1 week of insulin infusion, she was converted to basal-bolus regimen with subcutaneous regular insulin and NPH insulin which was technically challenging due to difficulty in dilution and marked glycaemic variability. At 2 months old, she was placed on continuous subcutaneous insulin infusion (Medtronic Minimed 780) with continuous glucose monitoring system.

The onset of NDM is less than 6 months-old and genetic testing is indicated for patients diagnosed before 9 months-old. Typical presentations are SGA, dehydration and hyperglycaemia. Bolus subcutaneous insulin is not ideal in NDM because of frequent feeding, limited subcutaneous fat, reduced insulin requirement, and huge fluctuations in glycaemic levels regardless of the type of insulin used. The successful use of insulin pump has been described in NDM.