Paediatrics E-Poster Presentation

EP_P001

THYROID HORMONE RESISTANCE SYNDROME (THR): A CASE REPORT

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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

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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.

CONCLUSION
Genetic mutations in children with NDM are common. Insulin remains the mainstay of therapy in INS-gene mutation. Genetic testing should be done to facilitate management.

EP_P003

ZOLEDRONIC ACID THERAPY FOR MONO-OSTOTIC LANGERHANS CELL HISTIOCYTOSIS: A CASE REPORT

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INTRODUCTION/BACKGROUND
Langerhans cell histiocytosis (LCH) is a rare disease. It can affect any organ in the body but is primarily characterized by osteolytic bony lesions. Skeletal LCH may range from a unifocal, self-limiting, asymptomatic lesion to severe, painful, destructive lesions that are prone to pathological fractures. Treatment decisions are individualized according to location, size, surgical accessibility, and functional impairment. Hence, there is no standard of care at the moment. Zoledronic acid (ZA) has been used in some neoplastic bone conditions to slow down the progression and reduce the bone pain.

CASE
We report a 6-year-old male with unifocal bony LCH at the left tibia who responded well to ZA. He presented at 4 years old with limping and was subsequently not ambulatory due to severe pain. He had a tender swollen left shin without skin changes.

X-ray showed a poorly defined 4.7 x 1.1 cm permeative lytic lesion in the medullary cavity of the midshaft of the left tibia with endosteal thinning. Subsequent MRI and isotope bone scans (Tc-99m MDP) confirmed a suspicious primary bone malignant lesion. A bone biopsy showed a neoplastic proliferation of histiocytoid cells with strong diffuse positivity for CD 1a, which was in keeping with LCH.

This confirmed a symptomatic mono-ostotic LCH with significant cortical destruction that was at risk of fracture. ZA was initiated after careful evaluation. His pain completely resolved with a return of function six weeks after the first dose of ZA. He received 4 doses of ZA in total and demonstrated radiographic evidence of regression and remained in remission.

CONCLUSION
This case demonstrates the potential role of ZA therapy as the first line treatment for mono-ostotic LCH stabilisation and symptomatic control.

EP_P004

CASE REPORT: MUCOLIPIDOSIS BONE DISEASE RESEMBLING NEONATAL RICKETS

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INTRODUCTION/BACKGROUND
Mucolipidosis II (I-cell disease) is a rare genetic metabolic disorder of lysosomal metabolism with a combined frequency of 1:422,000, that is characterized by coarse facial features, disproportionate short stature, hyperplastic gums, organomegaly, and retarded psychomotor development. These physical changes however require time to develop and are not apparent at birth. Opportunistically, neonates with I-cell disease are diagnosed after revealing typical spine changes in skeletal survey. Unlike nutritional rickets, their underlying cause and course of the bone disease are different.

CASE
We report a premature 34-week-old male with a birth weight of 1.6 kg. He was born in an ambulance while his mother was being transferred from a district hospital. He required cardiopulmonary resuscitation at birth and was admitted to the neonatal intensive care unit where he stayed for another 6 months due to respiratory reason. During his 1st to 4th months of life, he had markedly raised ALP level (>1000 IU/L), together with severe hyperparathyroidism that gradually resolved with time. He had radiographic changes resembling rickets which persisted and progressed to "chronic osteitis fibrosa cystica". His serum calcium, phosphate, and 25(OH)D_3 levels had always been normal. He never had a fracture. His diagnosis was later confirmed by marked elevation of plasma b-hexosaminidase, b-mannosidase and a-mannosidase. He was discharged home with tracheostomy and CPAP. He continues to survive at 11 months old by the time of this report.

CONCLUSION
Mucolipidosis osteodystrophy could resemble nutritional rickets during the neonatal period but it is not related to either deficient vitamin D or minerals. Failure to respond to conventional treatment (vitamin D and calcium or phosphate), should prompt for a search for other causes of increased bone turnover. In I-cell disease,