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SDHB Mutation in a Child with Paraganglioma: A Case Report

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INTRODUCTION

Paediatric paraganglioma (PGL), a rare endocrine tumor, originating from neural crest along sympathetic or parasympathetic chain, is highly associated with the syndromic presentation of MEN, NF1 or von Hippel-Lindau, and the germline mutations that affect succinate dehydrogenase gene (SDH).

RESULTS

We described a 12-year-old girl who had insidious onset of heat intolerance, diaphoresis and palpitation for 1 year. Her symptoms were initially transient but became intense and associated with episodic attacks of abdominal pain and vomiting. She was hyperglycemic during admission with weight loss of 3 kg, polyuria and polydipsia for 2 months. Clinical examination revealed hypertension and a vague mass palpable over right lumbar region. CT abdomen showed a right retroperitoneal enhancing mass with vascular displacement and compression. Urine catecholamine revealed raised norepinephrine 1658.0 ug/day (15-80). She was commenced on oral phenoxybenzamine with addition of oral metoprolol during the 2nd week. Her blood sugar was controlled with SC insulin. She had liberal intake of oral fluid and oral NaCl supplement up to 4.5 gram/day. PET scan (DOTANOC) showed somatostatin receptor avid primary disease with no distant metastasis. Laparotomy was performed and a well-circumscribed mass measuring 6 cm x 4 cm, which was not adherent to the kidney vessels, was resected successfully. Transient hypertensive crisis occurred during surgical resection and responded to bolus intravenous sodium nitroprusside. Postoperatively, she did not require SC insulin and antihypertensive medications. Histopathological findings are consistent with paraganglioma. Her genetic results showed heterozygous mutation in SDHB gene for a variant designated c136C>T.

CONCLUSION

All paediatric paraganglioma should ideally have mutation analysis. Higher risk of metastatic disease and tumor recurrence are associated with SDHB mutation. Regular follow-up and monitoring of plasma metanephrine and normetanephrine or 24 hour urinary fractionated metanephrines are warranted.

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Hypertriglyceridemia Thalassemia Syndrome in an Infant Presented with Anemia: A Case Report

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INTRODUCTION

Hypertriglyceridemia Thalassemia Syndrome is a rare condition with few reported cases. In here, we reported an interesting case of Hypertriglyceridemia Thalassaemia Syndrome in a young infant presented to our centre.

CASE

A 1-year-old Malay girl presented with poor growth, progressive pallor and lethargy for the past six months. She had no significant family history of blood or lipid disorder. On examination, she is small for her age, pale and icteric. Liver and spleen were palpable 4cm below costal margin respectively. She has no lymphadenopathy or xanthomas. Laboratory investigations showed low haemoglobin level (Hb 7.2 g/dL) but normal platelet count (375 x 10^9/L), and white cell count (10 x 10^9/L). She had raised reticulocytes count (11%) and lactate dehydrogenase (LDH 532U/L). Direct coombs test was negative. Peripheral blood smear revealed hypochromic microcytic red blood cells with polychromasia and severe anis poikilocytosis. Her serum was grossly lipidemic after centrifugation. Lipid profile showed raised triglyceride level (TG 9.05 mmol/L) but other lipid parameters are in normal range (total cholesterol 2.85 mmol/L, HDL-c 0.26 mmol/L, LDL -1.25 mmol/L). Her infective status is negative and her thyroid function test is normal. Other acquired causes of hypertriglyceridemia are also excluded. She was transfused with total 40 cc/kg pack cells and her Hb raised to 12 g/dl and no more lipidemic serum sample. Hb analysis revealed HbE- Beta thalassemia. Her mother`s blood showed Hb-E trait with normal lipid profile. She defaulted follow up until 4 months later she presented to our care again with similar presentation.

CONCLUSION

Hypertriglyceridemia thalassemia is rare reported association and regular blood transfusion usually resolved the high TG level. Early recognition is essential to deal with complications such as acute pancreatitis or increased coronary risk.