PP-92

A Case Report on Congenital Hyperinsulinism due to ABCC8 Gene Mutation

https://doi.org/10.15605/jafes.034.S104

Pian T, Jeanne SL Wong, Janet YH Hong, Nalini MS, Annie L, Sze Teik T, Arliena A, Haiza H, Fuziah MZ
Paediatric Endocrine Unit, Hospital Putrajaya, Malaysia

INTRODUCTION

Congenital Hyperinsulinism (CHI) is the most common cause of persistent hypoglycemia in infants. It is a disorder caused by dysregulation of insulin secretion from pancreatic β-cells. There are 12 different gene mutations identified at this moment with ABCC8 and KCNJ11 genes mutation encode SUR 1 and KIR 6.2 subunit of the ATP-sensitive potassium channel (KATP) being the most common. Children with homozygous ABCC8 and KCNJ 11 mutations usually do not respond to diazoxide.

CASE

We report a female Indonesian infant with autosomal recessive CHI secondary to ABCC8 gene mutation.

She was born term with birth weight of 4080 gram without history of maternal gestational diabetes. She developed symptomatic hypoglycemia at 10 hours of life with random blood glucose of 1.7 mmol/L. She needed a high glucose delivery rate up to 18 mg/kg/min and glucagon infusion to maintain normoglycemia. Critical blood sampling confirmed hyperinsulinism with an elevated serum insulin level of 18.1 U/ml at a random plasma glucose of 1.6 mmol/L. She had recurrent hypoglycemia despite an optimal dose of diazoxide. Subsequently, she was transferred to our center at the age of 2 months and started on subcutaneous octreotide injection four times daily; needing up to 35 mcg/kg/day to maintain blood glucose levels above 4 mmol/L. Genetic testing revealed a homozygous mutation in the ABCC8 gene which is known to be associated with poor response to oral diazoxide. Recessive ABCC8 mutation is associated with diffuse form of CHI hence optimizing medical treatment will be the preferred option before deciding for near total pancreatectomy.

CONCLUSION

This case illustrates a severe neonatal hypoglycemia unresponsive to diazoxide, where genetic confirmation helps to prognosticate the outcome and to plan for treatment strategy. It is vital to make an early diagnosis of CHI and initiate appropriate management to prevent hypoglycemia related permanent neurological damage.

PP-93

Thyroid Abscess in Children: A Case Series

https://doi.org/10.15605/jafes.034.S105

Sook Weih L,[1,2] Mazidah N,[1,3] Sheena D,[2] Azriyanti AZ,[3] Muhammad Yazid J,[3] Noor Shafina MN[1,4]
[1]*Department of Paediatrics, Faculty of Medicine Universiti Teknologi MARA (UiTM), Sungai Buloh Campus, Selangor, Malaysia*
[2]*Department of Paediatrics, Hospital Sungai Buloh, Sungai Buloh, Selangor, Malaysia*
[3]*Department of Paediatrics, Faculty of Medicine, University Malaya, Kuala Lumpur, Malaysia*
[4]*Institute for Pathology, Laboratory and Forensic Medicine (I-PPerForM), Universiti Teknologi MARA (UiTM), Sungai Buloh, Selangor, Malaysia*

INTRODUCTION

Thyroid gland has low risk of infection due to good vascular supply, lymphatic drainage, high iodine content and enveloping thick fibrous tissue. Thyroid abscess is rarely found in children. We report two cases of thyroid abscess.

CASE 1

A 2-month-old girl presented with painless neck swelling for 2 days. There was no history of fever, respiratory or thyroid symptoms. No history of maternal thyroid disease. Examination revealed firm non-tender midline neck swelling measuring 3 cm x 3 cm, normal overlying skin and accompanying cervical lymph nodes. White cells were raised $19.9x10^9$ with increased ESR 97 mm/hr and CRP 6.58 mg/dL [NR <3 mg/dL] with normal thyroid function test (TFT). Thyroid autoantibodies were not performed. Ultrasound revealed well-encapsulated heterogenous mass from left lobe measuring 2.2 cm x 3.9 cm x 4.0 cm causing deviation of trachea medially and left CCA IJV laterally. Aspirated pus yielded Klebsiellae pneumoniae and Enterobacteriae cloacae. TB culture was negative. She received intravenous cloxacillin and cefuroxime and discharged well.

CASE 2

A 7-year-old boy presented with one-week history of neck pain, fever and neck swelling. There was no trauma or flu-like symptom. Examination revealed 7 cm x 4 cm swelling at anterior, right neck with multiple shotty cervical lymph nodes. TFT was normal with negative inflammatory markers, anti-TPO and anti-TG antibodies. Ultrasound showed heterogenous right lobe measuring 2.8 cm x 3 cm x 5.1 cm with multiple right cervical lymph nodes (largest 0.6 cm). FNAC showed inflammation with perivasculitis, vasculitis with no granuloma or malignant changes. Patient