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A Case Report on Congenital Hyperinsulinism due to ABCC8 Gene Mutation

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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.

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Thyroid Abscess in Children: A Case Series

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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

developed purulent discharge after 2 weeks but afebrile. Well-circumscribed swelling was noted at the right anterior neck. Purulent discharge was aspirated with negative culture. Histopathology showed granulation tissue with infiltration by inflammatory cells with micro abscesses. Patient was given co-amoxiclav and discharged well.

CONCLUSION

Thyroid abscess must be considered although very rare in children. Intensive and appropriate treatment is necessary to prevent recurrence. Anatomical abnormalities like pyriform sinus fistula must be considered especially with atypical organisms or recurrent presentation.

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Use of Thiazide Diuretics in the Management of Central Diabetes Insipidus in a Neonate

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INTRODUCTION

The treatment of central diabetes insipidus (DI) with Desmopressin in the neonatal period is challenging because of the significant risk of hyponatremia. The fixed anti-diuresis action of Desmopressin and the obligate high fluid intake with milk feeds may lead to considerable risk of water intoxication and hyponatremia in neonates. Few case reports described the use of thiazide diuretics for treatment of central DI in infancy which was switched to Desmopressin later in life.

METHODOLOGY

We present a case of a premature female baby with midline defect, central DI and poor weight gain.

RESULTS

She was started with oral hydrochlorothiazide dose of 0.5 mg per kg per dose two times daily. Throughout the hospital stay, the dose was adjusted to 0.48 mg per kg per dose twice daily to achieve a stabilized serum sodium values ranging between 140-145 mmol/L. She has no obvious complications of hyponatremia. She was thriving well during follow up.

CONCLUSION

Oral thiazide diuretics is an alternative treatment of central DI in neonates. It is effective to achieve adequate control of DI without wide serum sodium fluctuations.

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Adrenocortical Carcinoma Presenting as Malignant Hypertension with Intracranial Bleed

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INTRODUCTION

Adrenocortical carcinomas are rare tumours with a bimodal distribution, peaking at the age of less than 5 years and also around the 5th decade. In children, virilisation is the most common presentation while Cushing's syndrome and hyperaldosteronism are less frequent.

CASE

We present a 6-month-old girl of Bangladeshi descent who presented at the age of 2 months old with status epilepticus following a trivial fall. She sustained a left intraventricular bleed with right front parietotemporal subarachnoid bleed. She underwent a right ventriculoperitoneal shunt insertion for obstructive hydrocephalus. Post operatively, she was noted to have recalcitrant hypertension with poor response to three antihypertensive therapy i.e. oral nifedipine, prazosin and captopril. During her hospitalisation, she developed rapid weight gain with development of facial acne and increasing facial, pubic and axillary hair.

Hormonal investigations revealed elevated testosterone of 52.05 nmol/L, elevated DHEA of >27.1 µmol/L and elevated 17 hydroxyprogesterone of >60.6 nmol/L. Her morning (8am) cortisol was 1494 nmol/L while 12 midnight cortisol was 1493 nmol/L. A CT abdomen revealed a large right suprarenal mass measuring 5.5 cm x 6.4 cm x 6.6 cm. The tumour (9 cm x 8 cm) was removed completely at five months old, however intraoperatively it was noted to have capsular breach and tumour spillage. Histopathological examination confirmed the diagnosis of high-grade adrenocortical carcinoma. A repeat CT abdomen done two weeks post-operative, unfortunately revealed tumour recurrence measuring 3.9 cm x 4.5 cm x 4.8 cm at the subhepatic region. Hence chemotherapy (Cisplatin/Etoposide/Doxorubicin) was initiated with addition of Mitotane. Postoperatively, her hypertension is gradually resolving within six weeks after surgery.

CONCLUSION

Invasive adenocarcinoma carries a poor prognosis. Early evaluation for this condition is vital in the presence of hypertension and virilisation in young children.