intrapartum treatment (EXIT procedure) at 38 weeks and 4 days. The infant was intubated and ventilated for respiratory distress. Newborn examination showed an anterior neck mass measuring 2 x 2 cm from midline to the left, with otherwise unremarkable systemic examination. Postnatal computed tomography of the neck revealed massive goitre causing airway compression and oesophageal narrowing from the oropharynx until the thoracic inlet. Laboratory studies supported the diagnosis of congenital hypothyroidism (TSH 37.41 μIU/mL) with possible thyroid dyshormogenesis. Treatment was initiated with oral levothyroxine 50 mcg daily. Serial ultrasound imaging showed a gradual reduction with resolved mass effect and airway compression.

CONCLUSION

Prompt diagnosis and meticulous thyroid replacement therapy led to significant regression of goitre to a more functional size. Rational intervals of clinical and biochemical evaluation are crucial to ensure optimum growth and neurodevelopmental outcomes.

EP_P007

INFANTILE HYPOCALCAEMIC SEIZURE AND VITAMIN D DEFICIENCY

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INTRODUCTION/BACKGROUND

Growing evidence suggests that vitamin D is essential for maternal and child health in many aspects. Nevertheless, a severe manifestation of vitamin D deficiency in the form of hypocalcaemic seizures continue to occur among Malaysian infants.

METHODOLOGY

A descriptive cross-sectional study was performed in the Paediatric Endocrinology Unit, Hospital Putrajaya. Records of all infants with hypercalcaemic seizures managed by our unit between January 2015 until April 2024 were retrieved from the electronic database system. Causes of hypercalcaemic seizure among this group of patients were identified. Further clinical, biochemical and hormonal results to assess the calcium-vitamin D-PTH axis were analysed.

CASE

A total of 24 patients were treated for hypercalcaemic seizures during the study period. Sixteen patients were male. Majority (75%) of the patients had hypercalcaemic seizures secondary to vitamin D deficiency, while 25% had hypoparathyroidism.

Among the group of hypercalcaemic seizures secondary to vitamin D deficiency, the median age of presentation was 8 weeks. Their mean corrected calcium, phosphorus, magnesium and ALP on presentation were 1.4 mmol/L, 2.35 mmol/L, 0.73 mmol/L and 690 U/L respectively. Mean iPTH and vitamin D levels of the patients were 19 pmol/L and 16.4 nmol/L, respectively. Maternal vitamin D levels were available for 7 mothers, showing a low mean value of 28.7 nmol/L.

All patients with hypoparathyroidism in this study had concomitant vitamin D deficiency or insufficiency. The median age of presentation was 3.5 weeks. Their mean corrected calcium, phosphorus, magnesium and ALP upon presentation were 1.64 mmol/L, 2.80 mmol/L, 0.64 mmol/L and 384 U/L, respectively. Mean iPTH and vitamin D levels were 2.3 pmol/L and 41.7 nmol/L, respectively.

CONCLUSION

Vitamin D deficiency or insufficiency was present in all patients in our study population. Vitamin D deficiency remains the predominant cause of hypocalcaemic seizure. Thus, vitamin D supplementation for all pregnant women should be encouraged as part of routine care. All infants during the first year of life should be encouraged to receive an oral vitamin D supplementation.

EP_P008

MALIGNANT GONADAL TUMOUR IN TRISOMY 21 WITH COMPLETE SEX REVERSAL

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INTRODUCTION/BACKGROUND

Trisomy 21 is a chromosomal disorder with a high incidence worldwide. It is associated with characteristic physical features, delay in development and some congenital organ defects. However, disorder of sex development (DSD) is not usually seen in patients with Down Syndrome.

CASE

A 14-year-old phenotypically female with Down syndrome presented with a two-month history of progressive abdominal swelling, constipation and weight loss. At birth, typical Down Syndrome facial features were present and the female gender was assigned. The karyotype result of 86 cells showed 47XY, +21 with +SRY gene via FISH study.

Pelvic ultrasound at 2 months showed the presence of a uterus and 2 ovaries. A β-hCG stimulation test performed exhibited a suboptimal response: serum testosterone increased to 1.2 nmol/L from a baseline of 0.7 nmol/L. The patient subsequently failed to turn up after a planned diagnostic laparoscopy was cancelled.

At the current presentation, the patient was short and thin. Female pubertal changes were present. The abdomen was distended with a firm rounded palpable mass measuring 20 x 15 cm. There were no findings of clitoromegaly nor palpable gonads.

Primary gonadal failure was evident from high serum gonadotropins and disproportionately low levels of oestrogen and testosterone. Abdomen CT showed a highly vascularised mass arising from the anterior abdomen with multiple septations of mixed cystic and solid components with calcifications; the uterus and two ovaries were seen. Laparotomy revealed a huge mass measuring 15 cm x 16 cm x 6 cm and two gonad-like structures with bridging Müllerian structures and abnormal-looking lymph nodes. Histopathology revealed dysgerminoma and gonadoblastoma of the huge mass and the gonads and metastatic changes in the lymph nodes. A PET scan showed metastasis to the right lung. The patient underwent chemotherapy subsequently.

CONCLUSION

DSD in Down Syndrome with Y chromosome is at high risk of gonadal tumour. Prevention and early detection are possible with the continuation of surveillance and meticulous assessments.

EP_P009

CO-INCIDENTAL FINDING OF SUPRATENTORIAL EPENDYMOMA IN PATIENT WITH GRAVES' DISEASE

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INTRODUCTION

Ependymomas are rare primary tumours of the cranial nervous system in children, which can be classified into supratentorial, infratentorial and spinal cord. They may present with neurological deficits or an increase in intracranial pressure symptoms based on their anatomical sites. Their symptoms may overlap with Graves' disease. It is postulated that the occurrence of Graves' disease might be due to alterations in the immunological response involving the hypothalamus-pituitary-thyroid axis that results in the formation of TSH antibodies.

CASE

A 10-year-old male who was previously well presented with lethargy, loss of appetite, recurrent vomiting and loss of weight for the past month. Clinically, he had bilateral exophthalmos, no ophthalmoplegia or lid lag, and a pulse rate of 120 beats/min.

His thyroid function test showed overt hyperthyroidism (TSH 0.60 m IU/L, T4 24.70 pmol/L). However, his thyrotropin receptor antibodies are still pending. Thyroid ultrasound was consistent with thyroiditis. He was started on thyroid storm treatment and his condition improved.

He presented again with reduced consciousness with a Glasgow coma scale of 10/15, unequal pupil and hyperreflexia of the left limbs. Urgent brain CT brain revealed a right cerebrum intra-axial tumour. He underwent tumour excision. Histopathology revealed a supratentorial ependymoma.

Post-operative Cranial MRI showed tumour size reduction measuring from 7.9 x 6.4 x 9.1 cm to a residual of 2.7 x 2.5 x 2.7 cm over the right parietal lobe. Carbimazole was continued and his clinical course was monitored.

CONCLUSION

This is a rare case of a co-incidental finding of supratentorial ependymoma with Graves' disease.

EP_P010

NEONATAL GOITER WITH AIRWAY AND OESOPHAGEAL COMPRESSION WHICH IMPROVED WITH L-THYROXINE AND A CONSERVATIVE APPROACH

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INTRODUCTION/BACKGROUND

Congenital goitre occurs rarely in neonates and it becomes an emergency when it causes significant airway compromise in newborns transitioning from foetal to postnatal life. neonatal goitre is closely related to thyroid status and could be either a transient or permanent genetic condition.

CASE

Foetal ultrasound in 2nd trimester detected an anterior neck mass with increased vascularity. At 38 weeks gestation,