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.