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,

a baby girl was delivered with a birth weight of 3.37 kg, length of 51 cm and OFC of 33 cm. She was intubated with ETT size 3.0 and ventilated in NICU. Examination showed a diffuse neck swelling measuring 3 x 3 cm. Ultrasound and CT scan showed an enlarged right thyroid lobe 3.5 x 2.7 x 4.2 cm (AP x W x CC) and a left thyroid lobe 3.1 x 2.8 x 4.3 cm (AP x W x CC) with no cystic component or calcification. Thyroid lobes extended up to the angle of the mandible and inferiorly until thoracic inlet with the airway and esophagus almost circumferentially encased and airway patency only maintained by ETT. Cord TSH 25.12 m IU/L, fT4 10.0 pmol/L, while Day 5 TSH 37.41 m IU/L, 6.47 pmol/L. ATPO, ATG & TSH receptor antibodies were negative. Her thyroglobulin level was low at 0.6 ng/ml (intact thyroid, 3.5-77.0). Her mother also reported onset of goitre following 1st trimester. She complained of tiredness and started taking Himalaya salt. Urine iodine results for patient and mother results were 322.7 ug/L and 221.3 ug/L, respectively (250 - 499, sufficient for pregnant mother). She was treated with L-thyroxine at day 5 of life at 50 mcg daily (15 mcg/kg) with serial ultrasound neck and flexible endoscopic assessment. She was successfully extubated by day 26 and discharged after 1 month. On follow-up, her goitre remained small with normal development and hearing. Due to cost constraint, genetic test was not pursued.

CONCLUSION

Neonatal goitre with hypothyroidism may result from maternal ingestion of antithyroid drugs or goitrogens, transplacental transfer of antithyroid antibodies, or thyroid dyshormonogenesis. Low thyroglobulin level with raised TSH and low fT4 suggest thyroglobulin synthetic defect.

EP_P011

A BOY WITH UNTREATED PANHYPOPITUITARISM: CASE REPORT

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INTRODUCTION/BACKGROUND

Hypopituitarism in childhood is a complex disorder with diverse clinical presentation which can either be congenital or acquired. Hormonal deficits can evolve over time leading to a significant impact on a child's growth.

We describe a case of untreated panhypopituitarism presenting as an adrenal crisis managed in a district hospital.

CASE

A 13-year-old Indonesian male with a background of panhypopituitarism post mature teratoma resection in January 2020 presented an adrenal crisis after he defaulted treatment for 2 years. On presentation, he had hypoglycaemia with shock requiring extensive fluid resuscitation and double inotropic support. Examination revealed weight and height below 3rd centile, pre-pubertal with thin eyebrows and depressed tendon reflexes. Height velocity was 4.2 cm/year for past 2 years. His random cortisol was extremely low (<1.5 nmol/L) with hypothyroidism. Intravenous stress dose hydrocortisone was initiated and his hemodynamic status improved over time. Oral thyroxine supplement was restarted and he required regular dose of sublingual desmopressin for diabetes insipidus.

CONCLUSION

Untreated panhypopituitarism has been reported in adult as late as 45 years old with significant impairment in cardiac function, growth and regression of sexual characteristic. With appropriate hormonal replacement, growth can be optimized and lifetime expectancy can be improved without long term sequelae.

EP_P012

DELAYED PRESENTATION OF PITUITARY TUMOUR WITH HYPOPITUITARISM

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INTRODUCTION/BACKGROUND

Hypopituitarism is a relatively uncommon disorder in the paediatric population, and its prevalence in children is not yet well established. This condition can be caused by any disease that affects the pituitary gland, stalk or hypothalamus. We describe a patient who presented late with short stature and hypopituitarism.

CASE

A 16-year-old male presented with short stature and delayed puberty. He denied any headaches or visual problems. He was born full-term with a birth weight of 3.2 kg. Developmental milestones were normal. At presentation, his height was 139 cm and his weight was 35 kg (both below 3rd percentile). Midparental height was 167.5 cm. His Tanner stage was 1 and testicular volume was 2 ml bilaterally. His visual field assessment was unremarkable. Hormonal analysis demonstrated