She has no other endocrinopathies. Her other comorbidities include coarctation of aorta, bicuspid aortic valve with severe aortic stenosis, post-balloon valvulotomy and coarctation repair. In view of her short stature, she was planned for GH therapy. Assessments pre-GH therapy revealed an incidental finding of central apnoea from polysomnography with an Apnoea-Hypopnea Index (AHI) of 22.5/H. This has led to MRI brain that revealed cerebellar tonsil descended 7 mm below the foramen magnum, consistent with Arnold-Chiari Type I malformation.

CONCLUSION

This case highlights the challenge of initiating GH therapy for a patient with Turner Syndrome and Arnold Chiari Type I malformation. Proper counselling with the patient and family is crucial to balance the harm and benefit of GH therapy. The decision to start GH therapy requires multidisciplinary management with close follow-up to monitor any complications and to avoid adverse events.

PA-P-05

46,XY DSD WITH HETEROZYGOUS MUTATION IN THE NR5A1 GENE: A CASE REPORT

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INTRODUCTION

Disorders of Sexual Development (DSD) is a rare disorder with a wide variable phenotype. These conditions occur rarely with a prevalence of about 1 per 5000 live births. Despite advances in genetic diagnostics, the underlying genetic cause in many of these patients remains elusive. One genetic cause for DSD, especially in individuals with 46,XY karyotype, is mutations in the NR5A1 (Nuclear receptor subfamily 5, group A, member 1) gene. NR5A1 encodes the transcription factor Steroidogenic Factor-1 (SF1) that plays a pivotal role in adrenal and gonadal development as well as in steroidogenesis. SF-1 is expressed in the bipotential gonad and regulates its differentiation towards testes and ovaries.

CASE

A 4-year-old child presented at birth with ambiguous genitalia. There was significant ambiguity of the genitalia presenting as micropenis (stretched penile length: 1.4 cm), perineal hypospadias, bifid scrotum with bilateral descended testis in the scrotum.

Initial investigations revealed chromosomal study of 46,XY, normal adrenal response on the ACTH stimulation test and an appropriate gonadotrophin surge during minipuberty. Beta HCG stimulation test revealed a poor testosterone response and the antimullerian hormone results were normal. Ultrasound of the pelvis and abdomen showed bilateral testes seen within the scrotal sacs and no Mullerian structures. Gender was subsequently decided as male following discussion with parents. Subsequently blood was sent for whole exome sequencing (WES) which revealed a heterozygous variant in NR5A1 gene.

CONCLUSION

In conclusion, we report a patient with 46,XY DSD with a heterozygous mutation in the NR5A1 gene. Patients with NR5A1 mutations regardless of phenotype at birth, may demonstrate considerable virilization at puberty. Therefore, it is important to consider gender assignment carefully in all patients.

PA-P-06

A CASE OF TRANSIENT DIABETES MELLITUS POST COVID-19 INDUCED DIABETES KETOACIDOSIS

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INTRODUCTION

The recent COVID-19 pandemic has highlighted the intimate connection between this novel virus and numerous endocrinopathies. Several studies reported increased incidence of paediatric diabetes particularly Type 1 diabetes mellitus presenting with diabetes ketoacidosis (DKA). We report a case of transient diabetes mellitus that presented in DKA secondary to COVID-19 infection.

CASE

A 15-year-old male with underlying pineal gland germinoma previously treated with surgery and cranial irradiation presented with a COVID-19 infection. He was treated for panhypopituitarism with thyroxine, hydrocortisone and DDAVP. Low dose growth hormone (GH) (0.016 mg/kg/day) was started after 5-years clinical remission. Pre-GH, his BMI was 24.2 kg/m^2 and the HbA1c was 4.9%. He complained of fever, respiratory distress, lethargy and reduced oral intake. At presentation, the plasma glucose was 52.2 mmol/L with high serum ketones of 7.6 mmol/L. Blood gas was acidotic (pH 7.25, bicarbonate 14.2 mmol/L). The HbA1c was 12.5% and the C-peptide was low. His COVID-19 PCR was positive. Fluid bolus was delivered, and he was managed as per DKA protocol. Stress dose hydrocortisone was given. After 12 hours he was transitioned to basal bolus subcutaneous insulin. After 1 month, he had recurrent hypoglycaemia prompting a reduction in the insulin doses and discontinuation after 2 months. The HbA1c and C-peptide level without insulin were 6.2% and 2.9 mg/ml respectively. His diabetes autoantibodies were negative.

CONCLUSION

COVID-19 infection is a potential trigger for development of new onset diabetes mellitus due to glucose dysregulation or autoantibody development. In our case, antibodies were negative and insulin dependency was temporary despite classically presenting with DKA. Long term follow up is required to monitor his glycaemic status.

PA-P-07

CENTRAL CONGENITAL HYPOTHYROIDISM IN AN INFANT OF A MOTHER WITH GRAVES' DISEASE

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INTRODUCTION

We report a case of central congenital hypothyroidism in a preterm baby born to a mother with hyperthyroidism.

CASE

The mother has been on treatment for hyperthyroidism since 2018. She developed thyrotoxicosis at 30 weeks pregnancy requiring Lugol's iodine, carbimazole and propranolol. She subsequently developed pre-eclampsia and went into labour. Her Thyroid Stimulating Hormone (TSH) receptor antibody levels were high, while anti-peroxidase and anti-thyroglobulin antibodies were negative. The child was born at 31 weeks gestation.

Initially the cord TSH level was 0.021 mIU/L. Subsequently, regular thyroid function tests continued to show very low TSH with normal T4. At one month of life, the T4 levels were low with persistent very low TSH and therefore the child was started on L-thyroxine. TSH receptor antibodies were positive. He was noted to have constipation and an umbilical hernia during this review. The diagnosis of central congenital hypothyroidism (CCH) was made, and the child was started on L-thyroxine. After initiation of therapy, T4 levels have normalised.

CCH is a rare condition with prevalence of 1 in 180 000 children. The risk is significantly increased in infants born to mothers with Graves' disease. In Graves' disease, patients may have TSH-blocking antibodies that bind to TSH receptors but do not initiate intracellular signaling, resulting in hypothyroidism. These antibodies can freely cross the placental, especially during the second and third trimester. Fetal thyroid matures functionally at around 25 weeks of gestation and because of this the hypothalamic-pituitary-thyroid axis can be affected in utero or postnatally.

CONCLUSION

This case highlights the importance of monitoring T4 and TSH levels in infants born to women with Graves' disease.