RESULTS

A male neonate was born prematurely at 31-weeks gestation via emergency lower segment caesarean for fetal distress with a birth weight of 1.7 kg. He was admitted to the neonatal intensive care unit due to respiratory distress requiring non-invasive ventilation. Newborn examination revealed hepatosplenomegaly with conjugated hyperbilirubinemia, hence, he was empirically treated for congenital infections. At 72 hours, the patient developed tachycardia and further work up resulted in suppressed cTSH. At that time mother was incidentally noted to have features of clinical Graves' disease which was confirmed by 2 thyroid function tests and positive TSH receptor antibody. Further radiological assessment of the newborn revealed periportal fibrosis and pulmonary hypertension. He was commenced on carbimazole and short course of prednisolone which resulted in resolution of hyperthyroidism, pulmonary hypertension, periportal fibrosis and thrombocytopenia.

CONCLUSION

Pulmonary hypertension and periportal fibrosis are rare clinical manifestations of neonatal Graves' disease which are reversible with resolution of hyperthyroidism.

PA-P-03

X-LINKED CONGENITAL ADRENAL HYPOPLASIA: A CASE REPORT

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INTRODUCTION

Adrenal hypoplasia congenita is a rare disease. It is characterized by primary adrenal insufficiency and/or hypogonadotropic hypogonadism (HH). Approximately 60% of affected males experience acute infantile onset while the remaining 40% have childhood onset. NR0B1/ DAX1 plays a pivotal role in the development and function of the adrenal and reproductive axes. Loss of NR0B1/ DAX1's inhibitory property due to NR0B1 mutations was demonstrated to be responsible for the pathology of X-linked adrenal hypoplasia congenita (AHC) and hypogonadotropic hypogonadism (HH).

CASE

We present a 15-year-old male who was initially referred to us at 1 year old when he presented with adrenal crisis. He was treated empirically with Hydrocortisone, Fludrocortisone, and sodium supplementation which was weaned off after infancy.

Synacthen test showed poor adrenal response with peak cortisol of <30 nmol/L, low 17OHP with ACTH levels of 9.7 pmol/L, suggestive of primary adrenal Insufficiency. During the course of follow-up he was noted to have delayed puberty. Physical examination showed no dysmorphism, normal blood pressure, prepubertal Tanner Staging with AH1PH1 and testicular volume of 2 mls bilaterally.

Stretched penile length was 5 cm with width of 1.5 cm (<10th centile). LHRH stimulation test confirmed HH, after which IM Testosterone was started. Genetic testing revealed a pathogenic mutation in the NR0B1 gene. (NP_000466.2:p. Ser175ValfsTer14) Hemizygous

CONCLUSION

In conclusion, we report a patient with adrenal hypoplasia congenita with novel mutation of NR0B1/ DAX-1 gene. Early diagnosis is important for long-term treatment in terms of endocrine and reproductive function and genetic counseling; the possibility of a NROB1/ DAX- 1 mutation must be considered in male patients with adrenal insufficiency.

PA-P-04

TURNER SYNDROME WITH ARNOLD CHIARI TYPE I MALFORMATION: A CASE REPORT

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INTRODUCTION

Turner Syndrome (TS) is a genetic disease caused by absence of one X chromosome, and is uncommonly linked with congenital CNS abnormalities. Arnold-Chiari Malformation is rarely associated with TS. Furthermore, there are limited reports available on the outcome of growth hormone (GH) therapy in this group of patients.

CASE

We described a 17-year-old female who was referred to us 3 years ago due to suspicion of TS in view of dysmorphism, short stature and primary amenorrhea.

Her karyotyping confirmed 45,X. Her height at presentation was 132 cm (- 4.98 SDS), weight 45.55 kg (-1.28 SDS), BMI of 26 kg/m^2 (+1.72 SDS), with MPH 153 cm. She was prepubertal with Tanner staging of A1, B1, PH1. In consistent with primary gonadal failure, her LH and FSH were elevated at 18.1 IU/L and 95 IU/L respectively with low Oestradiol <18.3 pmol/L. Her renal ultrasound was normal.

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.