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.