**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

*https://doi.org/10.15605/jafes.037.S2.83*

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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 (<10$^{th}$ 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

*https://doi.org/10.15605/jafes.037.S2.84*

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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.