CONCLUSION
It is crucial to ensure mothers with GD have early diagnosis and adequate monitoring during pregnancy to prevent neonatal complications. Infants of maternal GD should be monitored closely and at regular intervals, to detect alteration of thyroid function which is important for brain development.

EP_P038

THE ECLIPSE HAS PASSED

https://doi.org/10.15605/jafes.040.S1.268

Nurfarahiyah Nasuha Affandi and L Alexis Anand
Department of Paediatrics, Hospital Raja Perempuan Zainab II, Kota Bharu, Kelantan, Malaysia

INTRODUCTION
Primary adrenal insufficiency (PAI) can be misdiagnosed as other life-threatening conditions. Clinical signs of PAI are based on the deficiency of both glucocorticoids and mineralocorticoids. We report an infant with generalised hyperpigmentation and PAI.

CASE
A 10-month-old female, born via spontaneous vaginal delivery, with poor Apgar Score ($1^{1}3^{5}5^{10}$), was admitted for severe hypoxic-ischemic-injury (HIE). She had multiple episodes of seizures and required cooling therapy. Her parents are non-consanguineous. Clinically, she had generalised skin hyperpigmentation and normal female genitalia.

She had severe metabolic acidosis (pH: 6.7, $cHCO_3$: 8.3 mmol/L, lactate: 14.1 mmol/L). She developed an adrenal crisis at day 5 of life with lowest sodium: 125 mmol/L (134-142) and highest potassium: >7 mmol/L (3.5-5.6). The lowest blood glucose was 3.2 mmol/L.

Investigations at day 4 of life revealed low serum cortisol: 37.6 nmol/L (NV: 185-624), detectable testosterone: 0.9 nmol/L, normal 17-hydroxyprogesterone (17-OHP): 1.89 nmol/L (NV: <19.1), inappropriately low aldosterone: <103 pmol/L (NV: 471-4272) with high renin: >550 mU/L (NV: 4.00-89.00). The karyotype was 46 XX and the inborn error of metabolism study was non-diagnostic. Adrenocorticotropic hormone (ACTH) was normal at 5.16 pmol/L (NV: 1.60-13.90) but was done after initial doses of hydrocortisone. Pelvic ultrasonography (USG) showed Mullerian structures and cranialUSG was normal.

She was treated with stress dose of hydrocortisone 2.5 mg *qid* (45 mg/m^2/day), then weaned to oral hydrocortisone 1.5 mg *tid* (16 mg/m^2/day), fludrocortisone 150 mcg *od* and sodium chloride 0.5 grams *bid*. On follow-up, she had markedly reduced skin pigmentation. She had serum renin <1.80 mU/L and normal renal profiles. She was thriving with a weight of 7.4 kg (25-50th percentile), length of 65 cm (5-10th percentile) with appropriate developmental milestones.

CONCLUSION
Early diagnosis is crucial for effective management of PAI. Hyperpigmentation is a pathognomonic physical examination finding because ACTH shares the same affinity with α-melanocyte-stimulating hormone (MSH). Our patient's normal ACTH level was misleading due to hydrocortisone suppression.

EP_P039

WHEN THYROID STIMULATING HORMONE AND FREE THYROXINE MISMATCH: A CASE REPORT

https://doi.org/10.15605/jafes.040.S1.269

Yeap Cai Fong and Alexis Lordudass
Department of Paediatrics, Hospital Raja Perempuan Zainab II, Kota Bharu Kelantan, Malaysia

INTRODUCTION
Thyroid hormone resistance (THR) is characterized by lack of end-organ responsiveness to thyroid hormone with high serum free thyroxine (FT4) with inappropriately high thyroid stimulating hormone (TSH).

CASE
We report a 3-month-old male, born term via spontaneous vaginal delivery was referred to us at day 13 of life for inappropriately high TSH: 7.69 mIU/L (NV: 0.39-7.0) and high FT4: 57.5 mIU/L (NV: 8.7-16.2). His birth weight was 3.5 kg. Antenatally, his mother has no thyroid disorder. He is the only child of non-consanguineous parents.

The cord TSH was 22.01 mIU/L. At day 3 of life, TSH was 15 mIU/L and FT4 was insufficient. At day 23 of life, thyroid function test (TFT) revealed TSH: 6.29 mIU/L, FT4: 47.9 pmol/L and free triiodothyronine (FT3): 6.6 pmol/L (NV: 3.1-10.6). Clinically he had persistent left parieto-occipital swelling and tachycardia. He was started on carbimazole (0.1 mg/kg/day) at 2 months of life.

Thyroid autoantibodies were negative with anti-TSH receptor (TRAb) <0.8 IU/L (NV: <1.75), anti-thyroid peroxidase <9 IU/ml (NV: <35) and antithyroglobulin 14 IU/mL (NV: <115). Pituitary hormones taken on day 76 of life revealed prolactin 1,553.2 mIU/L (NV: 70.81-566.64) with mini puberty (testosterone: 4.3 nmol/L, FSH: 1.8 IU/L

Paediatrics E-Poster

and LH: 3.3 IU/L). At day 82 of life, prolactin was normal at 322 mIU/L and IGF-1 at 45.6 ng/ml (NV: 27-157). FBC, renal profile and liver function test were normal. Skull x-ray revealed no calcification and cranial ultrasound was normal. During follow-up, he has normal developmental milestones, weight: 5.3 kg (25th percentile), length: 55 cm (2nd percentile) and COH: 40 cm (75th percentile). Genetic confirmation testing is pending while awaiting funds. We plan to stop Carbimazole during follow-up.

CONCLUSION

We started carbimazole for tachycardia and suspicion of craniosynostosis. Treatment for THRB is not needed mostly, because the hyposensitivity to thyroid hormones seems to be adequately compensated by the increase in secretion of T4 and generation of T3.