hypopituitarism as validated by the following: LH <0.12 IU/L (1.8-11.8 IU/L) FSH 0.8 IU/L (3.03-8.08 IU/L) fasting morning testosterone 0.32 nmol/l (0.69- 26.16 nmol/l), IGF-1 38.7 ng/ml (226-903 ng/ml), morning serum cortisol 158 nmol/l (102-558 nmol/l) with inappropriately normal ACTH 4.37 pmol/L (1.6-13.9 pmol/L) fT4 7.74 pmol/L (11.4-17.6) TSH 2.08 m IU/L (0.47-3.41) prolactin 671.28 m IU/L (72.6-407.4). Synacten test revealed inadequate response with peak cortisol 184 nmol/l at 60 minutes. His bone age was delayed between 11 - 13 years. Magnetic resonance imaging of the pituitary gland revealed the presence of an enhancing lesion at the suprasellar region, at the centre of the optic chiasm abutting the proximal part measuring 1.0 x 1.2 x 1.1 cm (AP x W x CC). Differential diagnosis includes craniopharyngioma or pilocytic astrocytoma. He was replaced with glucocorticoid and levothyroxine while awaiting a parental decision regarding tumour excision.

CONCLUSION

Hypopituitarism can present in neonates, infants, children, and adolescents with multifactorial aetiologies. Timely diagnosis of this condition is crucial for effective intervention and management of affected children. The key to successful management of hypopituitarism lies in a high index of suspicion, coupled with increased awareness and appropriate hormone replacement therapy. Access to facilities for surgical intervention is essential for the survival and good prognosis of affected children.

EP_P013

INCREASING TRENDS OF CENTRAL PRECOCIOUS PUBERTY AMONG CHILDREN IN HOSPITAL PUTRAJAYA, 2004 TO 2024: A DESCRIPTIVE STUDY

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INTRODUCTION/BACKGROUND

Temporal trends worldwide demonstrate evidence of an earlier onset and progression of puberty worldwide. This study aims to describe the trends in Precocious Puberty among children in Hospital Putrajaya between 2004 to 2024. Data retrieved from the electronic database were reviewed. All patients diagnosed to have precocious puberty (i.e., onset of puberty before age 8 years for girls and 9 years for boys) in the Department of Paediatric Endocrinology of Hospital Putrajaya from January 2004 until April 2024 were included (n = 89). These patients were stratified according to the diagnosis; children diagnosed with Idiopathic Central Precocious Puberty (CPP) and normal variant puberty (i.e., Premature thelarche (PT) or Premature Adrenarche (PA).

CASE

Overall, a total of 89 children (86 (96.6%) girls; median [interquartile] age at diagnosis for boys, 7 [6;10] years; for girls, 7 [2;9] years) were registered with a diagnosis of CPP, PT, PA. Majority were Malay, 54 (60.7%), 25 (28.1%) were Chinese, 8 (9%) were Indian and 2 (2.2%) were Nigerians. Majority of the cases were idiopathic CPP, 81 (91%); with a median [interquartile] LH:FSH ratio of 1.4 [0.16;7.23]. The MRI findings show normal findings in 30 (35.3%), pituitary microadenoma in 29 (34.1%), and pineal gland cysts in 1 (1.12%). There was a general increase in the number of cases of CPP over time; between 2013 to 2018; 23 (25.8%), and a greater rise between 2019 to 2024; 61 (68.5%). Nearly half of the cohort had a body mass index (BMI) of overweight or obese 40 (41.3%); with median [interquartile] bone age, 4 [2;7].

CONCLUSION

This study demonstrated an increase in the number of patients with central precocious puberty over 20 years. We also demonstrated a possible association with an increased BMI and earlier onset of puberty in girls.

EP_P014

OBESITY IN TEMPLE SYNDROME

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INTRODUCTION/BACKGROUND

Temple syndrome is a rare imprinting disorder caused by a maternal uniparental disomy of chromosome, paternal deletion of 14q32 or isolated methylation defect of the MEG3-DMR. Review of the electronic medical records with salient clinical and investigations recorded.

CASE

MKA is an 8 years and 5 months old male who presented with central hypotonia with poor sucking at birth. He was delivered term at 2.62 kg. Antenatally, the mother had oligohydramnios. During clinic follow-up, MKA remained well but he remained obese with a BMI of more than 97th centile. He looked dysmorphic with plagiocephaly, narrow bifrontal diameter, almond-shaped eyes, downturned mouth, thin upper lip, thick earlobes, small hands and feet, left single palmar crease, pes planus and genu valgus.

Neurological examination revealed central hypotonia. He had failure to thrive during infancy where he was on orogastric feeding for 1 month, achieved brief normalisation at 1 year old, followed by hyperphagia and rapid weight gain since the age of 2 years old. He is a product of a non-consanguineous marriage. He also has isolated central hypothyroidism with left cerebral hemiatrophy.

Chromosomal study showed 46XY karyotype. A series of imaging including abdominal ultrasound, KUB, pelvis, cranium, echocardiogram and brain MRI - was normal. He was suspected to have Prader Willi Syndrome, but methylation test was normal. Genetics team co-managed and treated him for Temple Syndrome with maternal uniparental disomy 14 with Prader Willi Syndrome like phenotype.

CONCLUSION

Temple syndrome is largely manifested by physical defects necessitating obligatory supportive therapies early in the life of affected individuals. The syndrome has shown that a subset of patients develop obesity. BMIs increased along the reference curves in most patients. Endocrine anomalies are prevalent, with truncal obesity developing as early as 4 to 6 years.

EP_P015

IT'S MODY! NOT TYPE 2 DIABETES

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INTRODUCTION/BACKGROUND

MODY (maturity-onset diabetes of the young) is a rare form of diabetes which represents a clinically heterogeneous group of autosomal-dominant disorders caused by mutation in genes involved in beta cell development and insulin secretion. The classic phenotype of MODY includes nonketotic noninsulin-dependent diabetes with diagnosis before age 25 years and an affected parent. However, there is clinical overlap between MODY, type 1, and type 2 diabetes resulting in frequent misdiagnosis of MODY. Data collection was done by reviewing the electronic medical records of the selected patient and salient points were noted.

CASE

TSE, presented at the age of 9 years old with hyperglycaemia detected on routine monitoring. On presentation, she was asymptomatic and her investigations revealed nonketotic hyperglycaemia (RBS 10.5 mmol/L). Clinically there was no evidence of acanthosis nigricans and her BMI was within normal range. Further investigations revealed HbA1c of 8.6% with an abnormal OGTT (FBS: 8.9 mmol/L, 2 hours post: 20.97 mmol/L). Her pancreatic autoantibodies came back as all negative and had a high c-peptide level of 357.9 pmol/L. She was treated initially with oral metformin. On further history, the mother had early-onset diabetes and there was a strong family history of early-onset diabetes on the maternal side. Genetic testing by whole exome sequencing revealed a heterozygous variant in HNF1B. She was further investigated and renal function, lipid profile and ultrasound of the abdomen and kidneys were normal. She was subsequently transitioned to insulin therapy.

CONCLUSION

HNF1B has a wide phenotypic spectrum, and affected individuals may present with isolated renal disease, isolated diabetes, or both. This case highlights the importance of the precision medicine approach in MODY. Molecular genetic testing can identify the subtypes and has profound implications on diabetes treatment and prediction of future development of co-morbidities, allowing early preventive or supportive treatment.

EP_P016

HYPOPHOSPHATASIA, RARE BONE MINERALISATION DISORDER THAT MIMICS OSTEOGENESIS IMPERFECTA AT BIRTH

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INTRODUCTION/BACKGROUND

Hypophosphatasia is a genetic disorder characterized by impaired mineralization of bones and teeth that results in fractures and deformities. It affects less than 1:100,000 live births with different clinical spectrums. It is caused by loss-of-function mutations of ALPL gene that encodes tissue nonspecific alkaline phosphatase (TNSALP), leading to low activity of this enzyme that usually mediates the breakdown of inorganic pyrophosphate that blocks mineralization. We reported an infant who was initially admitted to NICU at birth.

CASE

Antenatally, the patient was detected with possible osteogenesis imperfecta and unilateral lung hypoplasia by a detailed foetal scan at 32 weeks gestation. His mother had GDM and underwent LSCS at 37 weeks 5 days with a weight of 2.42 kg. He was born with generalized bowing deformities of both upper and lower limbs, a broad