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

https://doi.org/10.15605/jafes.039.S1.230

Chiew Wah Loh,[1] Nalini M Selveindran,[2] Janet YH Hong,[2] Jayne AX Ong[2] Sasirekha K Morthy[2]

[1]*Department of Paediatrics Hospital Tunku Azizah, Kuala Lumpur, Malaysia*

[2]*Department of Paediatrics Hospital Putrajaya, Malaysia*

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

https://doi.org/10.15605/jafes.039.S1.231

Farah Nursyahirah Binti Nordin

Hospital Sultan Bahiyah, Malaysia

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