were (4.2cm vs 14.8cm)/year and (4cm vs 17.6cm)/year, (47 vs 329) Âμmol/L and (<15 vs 205) Âμmol/L, respectively. Hypoglycaemia, a common side effect of treatment was not reported. H's percentage body fat and muscle mass improved from 54.1% to 52% and 10.8kg to 12.4kg. One year treatment for both siblings' costs RM751,500 (USD 182,624.57).

CONCLUSION

LS is rare, yet a treatable cause for severe short stature. Albeit the exorbitant cost, treatment offers positive outlook.

PE-03

VITAMIN D DEFICIENCY RICKETS – A CASE SERIES: 'A TIP OF THE ICEBERG'

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INTRODUCTION

Vitamin D deficiency is the most common cause of rickets worldwide. In Malaysia, owing to the abundant sunlight exposure, it is believed to be uncommon, however it is likely to be under-reported. In addition, dietary calcium deficiency is an important cause of nutritional rickets in children above 1 year old in developing countries.

RESULTS

We report a case-series of 4 unrelated Malaysian children (aged between 1 to 3) born in Istanbul, Turkey presented with the classical clinical features and biochemical changes of rickets. They were all exclusively breast-fed during infancy with poor dietary calcium intake. Their workup showed normal Calcium, high Alkaline Phosphatase, low Vitamin D and high Parathyroid hormone levels, with radiographic changes of fraying and spraying of the wrist, consistent with Vitamin D Deficiency Rickets. Bowing of legs and widening of wrists joints also seen. Low maternal Vitamin D levels also support the diagnosis. All four children were treated with cholecalciferol (vitamin D3) and short-term calcium supplements. The children showed improvements in growth and normalization of biochemical parameters on follow-ups.

CONCLUSION

Meta-analysis in Turkish populations have shown high prevalence of Vitamin D deficiency leading to their national policy of vitamin D supplementation for infants. Our patients in this case series were neither immunized nor received the appropriate supplements during their stay in Istanbul, Turkey. Maternal vitamin D deficiency, restricted sunlight exposure due to clothing style and seasonal variations, poor dietary calcium intake were all the contributing factors to the nutritional rickets in our patients. Maternal Vitamin D levels could serve as an early indicator of possible deficiency if detected early. Awareness amongst our population was scarce, leading to a delay in seeking treatment/intervention. This case series aims to highlight the importance of vitamin D supplementation as well as ensuring adequate dietary calcium in prevention of nutritional rickets.

PE-04

A CLINICAL PROFILE OF MALAYSIAN PRE-SCHOOL CHILDREN WITH TYPE 1 DIABETES: OBSERVATIONS FROM A SINGLE CENTRE

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INTRODUCTION

Type 1 diabetes (T1DM) is the most common form of childhood diabetes in Malaysian children, the median age being 7.6 years. Worldwide, younger children are increasingly being diagnosed with T1DM, especially in the under 5's age group. Vague clinical symptoms may lead to a protracted presentation and increase the risk of severe complications. This study aims to report the clinical characteristics of Malaysian pre-school (<7 years) children with T1DM .

METHODOLOGY

A retrospective review of the demographic and clinical data on children < 7 years of age diagnosed with T1DM at the University of Malaya Medical Centre between January 1st 2010-Dec 31st 2019 was conducted.

RESULTS

There were 119 diagnoses of TIDM during the study period. Forty-percent (n=47) were pre-schoolers, mean age being 4.16 ± 1.85 years. Boys comprised 60% (n=28) and the Chinese ethnicity was predominant, 32% (n=15). DKA occurred in 79% (n=37) at presentation, of these 73% (n=27) were moderate-severe DKA. The mean Hba1c was 11.98 ± 1.95% and 80% (n=38) were positive for atleast 1 pancreatic antibody, GAD-65 70% (n=33) being the most frequent. PICU admission occurred in 47% (n=22) cases and the mean length of stay was 7.8 ± 2.7 days. The lowest rate of pre-school T1DM was seen in 2011, 20% (1/5 cases), and the highest in 2017, 63% (12/19 cases). Within the pre-schooler group, 43% (n=20) were misdiagnosed, respiratory infections being the most common 60% (n=12) misdiagnosis. Furthermore, 90% (n=18) in the misdiagnosed group presented in DKA, of which 35% (n=12) were severe.

CONCLUSION

T1DM is the most common form of childhood diabetes in Malaysia. Pre-school children <7 years of age with T1DM are often misdiagnosed, present in moderate-severe DKA and require PICU admission. Future multi-centre studies need to evaluate the risk factors contributing to these findings and the long-term outcomes in pre-schoolers with T1DM.

PE-05

GENDER REASSIGNMENT FOR LATE PRESENTATION OF 5-ALPHA-REDUCTASE DEFICIENCY: A CASE SERIES

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INTRODUCTION

Failure to convert testosterone into a more potent androgen-dihydrotestosterone(DHT) due to 5-Î ± -reductase (5Î ± R) deficiency results in incomplete masculinization of external genitalia in newborns with 46XY karyotype. The clinical spectrum can range from complete female appearance to nearly complete male phenotype. Affected individuals with female genital phenotype may present at puberty with virilization. It is a rare disorder, caused by an autosomal recessive inheritance.

RESULTS

We report 3 adolescents with severe 5Î ± R deficiency who were brought up as girls due to female genital phenotype at birth. No consanguinity reported. One had bilateral inguinal swelling with hernia repaired at infancy. They presented at post-pubertal age (17- to 21-year-old) with primary amenorrhea and virilization during puberty. Physical examination showed tall stature with obvious laryngeal prominence and absence of breast development. They had enlarged phallus with fused labioscrotal folds, hypospadias, and enlarged testis at the inguinal/scrotal region. They had male hair distribution. All 3 had been excellent in female sports and displayed incongruence in gender identities. Chromosomal analysis showed 46XY with SRY positive. The diagnosis of 5Î ± R deficiency was suspected based on adequate serum testosterone level at baseline and markedly elevated following human chorionic gonadotrophin(hCG) stimulation test with testosterone/DHT ratio >30. 17Î²-HSD was excluded due to the normal response of androstenedione. Molecular studies showed the genetic mutation in SRD5A2 which confirmed the diagnosis of 5Î ± R deficiency. Their gender role and gender identity were carefully evaluated. Gender was reassigned to male after seeking legal approval from local Islamic religious and federal authorities. The family and the affected adolescents were given psychosocial support. Multi-stages corrective surgery was performed by the urologist.

CONCLUSION

A careful neonatal assessment followed by adequate endocrine evaluation may reduce missed 5Î ± R deficiency during the neonatal period. Most importantly, improved public awareness of inconsistent secondary sexual characteristics from assigned gender could lead to early medical referral.