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MALE OR FEMALE? A CASE REPORT OF 17-BETA HYDROXYSTEROID DEHYDROGENASE 3 (17 β -HSD3) DEFICIENCY

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INTRODUCTION/BACKGROUND

17 β -HSD3 deficiency is a rare autosomal recessive cause of 46-XY Disorder of Sexual Development (DSD), occurring in approximately 1 in every 147,000 newborns. 17- β -HSD3 is encoded by the HSD17B3 gene (chromosome 9q22.32) and expressed in the testis where it converts the inactive steroid androstenedione to the active androgen testosterone. We report a case of DSD who presented with hoarseness of voice and primary amenorrhea.

CASE

A case of a 22-year-old who was raised as female presenting with primary amenorrhea at the age of 16, preceded by gradual hoarseness of voice since 12. Height was 158 cm. Pubertal assessment revealed Tanner stage 2 breast development, stage 3 axillary and stage 4 pubic hair distribution. Physical examination was notable of facial hair, male body habitus, a phallus measuring 4 cm in length and 2 cm in girth, bilaterally palpable inguinal mass and empty rugated labioscrotal folds. Laboratory parameters revealed low to normal testosterone, raised FSH, LH and mildly elevated estradiol levels, pelvic MRI revealed bilateral gonads seen in the inguinal region with a small corpus cavernosa, corpus spongiosum, seminal vesicles, prostate and no uterus or ovary. The child and her parents received psychological counselling and she desired to maintain a female gender identity. Genetic studies by whole exome sequencing revealed homozygous mutation of HSD17B3 gene.

CONCLUSION

46-XY DSD due to 17 β -HSD3 deficiency is a rare disorder that may prove a diagnostic conundrum in situations where specific endocrine panels and genetic testing are not readily available. In patients where the diagnosis is delayed there may be difficulty in gender assignment.

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VITAMIN D DEFICIENCIES: A PILOT STUDY IN OVERWEIGHT AND OBESE CHILDREN AND ADOLESCENTS

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INTRODUCTION

In the post COVID era, the prevalence of overweight and obesity in children and adolescents is increasing, leading to multiple obesity-related comorbidities. Despite Malaysia being a tropical country, the occurrence of vitamin D deficiency has been increasing. This study aimed to determine the incidence of vitamin D deficiencies in normal and obese/overweight groups.

METHODOLOGY

A total of 43 participants aged 7 to 17 years old were recruited from the Hospital Al-Sultan Abdullah, UiTM. During the hospital visit, anthropometric measurements and a questionnaire on dietary intake, sunlight exposure and average family income were collected. Total serum vitamin D levels were taken and analysed using the electrochemiluminescence assay.

RESULT

The mean age of the participants was 9.30 ± 1.89 years old. A total of 48.8% were overweight or obese. The overweight/obese group had a significantly higher body mass index (BMI) (29.01 ± 11.23 kg/m² vs 14.95 ± 1.77 kg/m², $p < 0.01$), weight (54.73 ± 22.7 kg vs 24.23 ± 7.24 kg, $p < 0.01$) and waist circumference (82.65 ± 23.27 cm vs 55.66 ± 5.10 cm,