CONCLUSION

CAIS is a genetic condition in which a child is genetically male but develops female sex characteristics. CAIS occurs when there is problem with one of the genes on the X chromosome at Xq11-12and code for protein with a molecular mass approximately 110kDa (androgen receptor gene). It governs how a developing fetus responds to androgen-hormones that bring about male characteristics. A child with CAIS has a genetic makeup of XY. Because the Y chromosome is present, the child is born with testis,, although the testes are undescended; but because of the defective gene on the X chromosome, other male characteristics don't develop, so the child resembles a female l**.** Most children with CAI are raised as female.

PE-19

SYMPTOMATIC HYPERCALCEMIA IN WILLIAMS SYNDROME

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INTRODUCTION

Williams syndrome is a multisystem contiguous gene deletion syndrome that presents with distinctive facial features, congenital heart disease, neurodevelopmental and behavioral deficits. Endocrine abnormalities such as diabetes and hypothyroidism are described in adults while hypercalcemia is mostly reported in infants and young children during first 2 years of life. Hypercalcemia in Williams syndrome is usually mild and asymptomatic resolving by the age of two years. It may be associated with hypercalciuria and to a lesser extent nephrocalcinosis, occurring in less than 5% of WS patients. Traditional treatment of hypercalcemia in children with William Syndrome consists of intravenous hydration, dietary restriction of calcium and vitamin D and in unresponsive cases, intravenous biphosphonate may be considered as second line treatment.

RESULTS

We report 2 children with Williams syndrome who presented with symptomatic hypercalcemia associated with nephrocalcinosis. Both these patients required hospital admission around the age of 2 years old and responded to intravenous hydration. We intend to highlight that symptomatic hypercalcemia in children with Williams syndrome is not uncommon and that their elevated serum calcium levels can respond to increased hydration via enteral and parenteral route in parallel with dietary restrictions.

CONCLUSION

In conclusion, close monitoring of serum calcium levels as well as parental education on symptoms of hypercalcemia and dietary advice is crucial in management of children with hypercalcemia in Williams syndrome.

PE-20

PAX 4 GENE MUTATION IN A 9-YEAR-OLD CHINESE BOY PRESENTING WITH DIABETIC KETOSIS

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INTRODUCTION

A 9-year-old Chinese boy presented with diabetic ketosis following one month history of polyuria, polydipsia and nocturnal enuresis. A strong family history of diabetes was present within his family pedigree whereby both his mother and maternal grandmother were diagnosed with type 2 diabetes at 15 years and 28 years old respectively. His 10-year old sister was also recently diagnosed with impaired glucose tolerance. The patient's body mass index (BMI) was normal at the 75th centile and there was absence of acanthosis nigricans. Anti-glutamic acid decarboxylase (GAD) and anti-islet tyrosine phosphatase 2 (IA2) were negative but anti-islet cell (ICA) was weakly positive. The patient was treated as type 1 diabetes with subcutaneous insulin therapy. Insulin treatment was withheld 1 month post diagnosis due to frequent hypoglycemia but subsequently restarted after 5 months post diagnosis, with gradual dose increment. The patient was able to maintain good glycemic control with insulin total daily dose of less than 0.5U/kg/day, alluding to the diagnosis of honeymoon period. The patient underwent genetic testing for MODY and was found to carry a heterozygous mutation of PAX4 gene, Exon 9, c.890G>A (p.Gly297Asp) of uncertain significance.

CONCLUSION

PAX4 mutation is a rare cause of MODY, initially reported in Thai patients. PAX4 mutations are associated with younger onset of type 2 diabetes, particularly in East Asians/Chinese. It is unclear if this child has type 1 DM or MODY due to PAX4 mutation. Further genetic testing of his family members is needed to determine the significance of this PAX4 variant and association with young onset diabetes. A more protracted follow up is needed to unveil this patient's diabetes progression and phenotype.