CONCLUSION
Partial ectopic posterior pituitary gland is a variant of ectopic posterior pituitary that is a rarely described imaging entity. Although there is a wide differential diagnosis for T1-hyperintensities (e.g., lipid-containing lesions, protein, metallic substances, methemoglobin and calcifications) on MRI, the diagnosis can be narrowed with the aid of additional MRI sequences and clinical manifestations.

PE-17

HYPOTHYROIDISM IN DOWN SYNDROME (DS) CHILDREN WITH CONGENITAL HEART DISEASE (CHD)

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INTRODUCTION
Down syndrome is the most common chromosomal disorder in liveborn infants, and is associated with thyroid dysfunction, estimated to occur in 4-8% of children. The reported incidence of congenital hypothyroidism (CHT) in children with Down syndrome is estimated 20 - 30 times higher than the incidence of CHT in the general population. We aim to investigate the prevalence of CHT in Down syndrome children with congenital heart disease (CHD) seen in our centre, as well as the aetiology of hypothyroidism in this group of paediatric patients.

METHODOLOGY
We performed a retrospective analysis of paediatric Down syndrome patients born between 1 January 1990 and 31 December 2020 who attended the paediatric cardiology clinic for routine review in the 3.25-year period between 1 September 2017 and 31 December 2020.

RESULTS
Children with DS accounted for 12.9% (n=62) of patients attending the paediatric cardiology clinic, with cyanotic CHD (cCHD) diagnosed in 29% (n=18) and acyanotic CHD (aCHD) in 71% (n=44).

The prevalence of hypothyroidism in our population of DS children with all types of CHD was 19.4% (n=12). However, among DS children with cCHD, the prevalence of hypothyroidism was 22.2% (n=4/18), which was higher than the prevalence of hypothyroidism in aCHD, 18.2% (n=8/44).

The mean age at presentation was 11.8 weeks (2 weeks – 32 weeks), and the mean TSH at presentation was 15.6 mIU/L (7.63 – 27.51 mIU/L).

All patients with suspected CHT (n=12) underwent thyroid scintigraphy at age 3. Six patients (50%) were confirmed to have permanent CHT: 42% (n=5) had thyroid dyshormonogenesis, and 8% (n=1) had thyroid dysgenesis. None had ectopic thyroid. Autoimmune hypothyroidism occurred in 8% (n=1). Subclinical hypothyroidism occurred in 42% (n=5).

CONCLUSION
The prevalence of CHT in DS children with CHD is 50% in this studied population. Children with DS tend to present later with subclinical hypothyroidism rather than high cord TSH. Routine screening in the first year of life is mandatory to detect thyroid abnormalities.

PE-18

CASE REPORT OF COMPLETE ANDROGEN INSENSITIVITY SYNDROME

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INTRODUCTION
Androgen insensitivity syndrome (AIS), also known as testicular feminization, encompasses a wide range of phenotypes that are caused by numerous different mutations in the androgen receptor gene. AIS is an X-linked recessive disorder that is classified as complete, partial, or mild based on the phenotypic presentation.

RESULTS
A 2 year and 10-month-old child, presented to private practice for inguinal hernia at 15 months age. Herniotomy was performed at 20 months age and there was intra operative finding of gonad in the inguinal canal. Histology confirmed male gonad. This child was raised as girl as external genitalia was phenotypically female, no clitoromegaly with 2 perineal opening. Karyotyping confirmed 46 XY with variant in AR gene:c.5A>G(p. Glu2GIy). Pelvic UTS showed presence of vagina, no uterus and ovaries visualised .FSH was 4.1 mU/mL, LH was 2.7 mU/mL, testerone was normal with value of <0.35, Antimüllerian hormone was elevated 164.2 pmol/L.

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.