revealed abnormal gonadal architecture in all cases. All intra-abdominal gonads demonstrated streak gonads or dysgenetic testicular tissue.

CONCLUSION

45,X/46,XY mosaicism demonstrates a broad phenotypic spectrum with frequent genital ambiguity, growth impairment, and abnormal gonadal development. The high prevalence of gonadal dysgenesis underscores the importance of careful surveillance and individualized multidisciplinary management guided by clinical, endocrine, and histopathological findings.

EP_P003

HEIGHT VELOCITY AND BODY MASS INDEX CHANGES DURING GONADOTROPIN-RELEASING HORMONE AGONIST THERAPY IN CHILDREN WITH CENTRAL PRECOCIOUS PUBERTY (CPP)

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INTRODUCTION

Precocious puberty is increasingly recognized in children. Effect of gonadotropin-releasing hormone agonist (GnRHa) on height velocity (HV) and body mass index (BMI) during treatment of Central Precocious Puberty (CPP) is not well-documented.

METHODOLOGY

Records of all patients treated with GnRH for CPP who attended the paediatric endocrine clinic over a 5-year period (2021–2025) [N1.1] were reviewed retrospectively for HV and BMI changes for 3 consecutive years. Basal luteinizing hormone levels ≥0.3 IU/L or stimulated levels ≥5 IU/L are considered diagnostic of CPP.

RESULTS

Twenty patients were reviewed and median age at the start of study was 7.8 years (4.3–10.3). Majority were female (n = 18, 90%) with median Tanner Stage 3 and of Malay origin (n = 18, 90%[N2.1]). Median chronological age was 7.8 years (4.3–10.3) with bone age 10.3 years (5.8–13.4).

Causes of CPP were idiopathic (n = 16, 80%), CNS lesion (n = 2, 10%) and poorly controlled congenital adrenal hyperplasia (n = 2, 10%). Nine (82%) of the 11 patients who had MRI brain done had normal findings.

After 1 year of treatment, the median HV SDS was −0.83 (range −2.45 to 2.42), and BMI SDS was 0.45 (−1.94 to 2.15). After 2 years (n = 12), HV SDS declined to −1.3 (−3.44 to 0.69), with BMI SDS of 0.13 (−1.16 to 1.79). After 3 years (n = 9), HV SDS declined further to −1.51 (−3.52 to 3.13), and BMI SDS was 0.16 (−0.12 to 1.25).

After 1 year of treatment, 20% (n = 4) of patients were overweight and 10% (n = 2) were obese. By 3 years, 22% (n = 2) remained overweight, and none were obese.

CONCLUSION

Premature growth plate senescence induced by prior estrogen exposure may lead to HV decline. Although obesity is a risk factor for the onset of CPP, whether GnRHa treatment directly increases BMI remains controversial as our cohort's BMI change reveals otherwise.

EP_P004

A SPECTRUM OF THYROID DYSFUNCTION IN CHILDREN WITH DOWN SYNDROME: A MALAYSIAN TERTIARY CENTRE EXPERIENCE

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INTRODUCTION

Endocrine abnormalities are frequently observed in children with Down syndrome, with thyroid dysfunction being the most common. The distribution and patterns of thyroid disorders vary between populations.

METHODOLOGY

Retrospective data were collected from the electronic medical records of children with Down syndrome who attended the paediatric clinic at University Malaya Medical Centre from the year 2000 to 2025. Data were analyzed to determine the frequency and distribution of thyroid disorders in this cohort.

RESULTS

The study included 57 patients, the majority of whom were identified through routine screening (n = 56, 98.2%). Only one child (1.8%) was diagnosed following a symptomatic presentation of diarrhea. Within the cohort, 51 patients were diagnosed with hypothyroidism and six with hyperthyroidism.

Among those with hypothyroidism, the mean TSH was 22.5 mIU/L, with a median age at presentation of 61 days (IQR: 19–211.5). Notably, 25% of the cohort presented within the

first 19 days, while the rest presented after 7 months of age. Of these, 24 cases (47.1%) were transient, with medications successfully discontinued at a mean age of 4.0 ± 2.13 years. Only 26 patients underwent thyroid scan, which was normal except for one case of thyroid agenesis. Imaging was not performed in others due to early discontinuation of therapy or loss to follow-up.

All hyperthyroid patients had autoimmune thyroid disease (4 Graves' disease, 2 Hashimoto's thyroiditis) with positive autoantibodies. The median age at presentation was 5.4 years (IQR: 2.3–10.2).

CONCLUSION

Screening for thyroid dysfunction successfully identified almost all cases in patients with Down syndrome. Notably, patients which hyperthyroidism in this cohort had co-occurring autoimmune thyroid dysfunction and demonstrated a significantly earlier age of presentation compared to the general paediatric population.

EP_P005

PREVALENCE OF HYPOTHYROIDISM AND GROWTH OUTCOMES AMONG PATIENTS WITH DOWN SYNDROME

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INTRODUCTION

Children with Down syndrome (DS) have a higher prevalence of hypothyroidism compared to the general population, and both conditions are linked to impaired growth. This study aimed to determine the prevalence and subtypes of hypothyroidism in DS and to compare growth outcomes with controls from general population at 2 years of age.

METHODOLOGY

A retrospective review was conducted on 248 children with DS (aged 2–18 years) followed at Hospital Pakar Universiti Sains Malaysia from 2020 to 2025. Growth outcomes were analyzed in a subgroup of 41 DS children with hypothyroidism compared to 46 controls. Growth was compared using standard CDC charts for and z -scores calculated with PediTools (CDC 2–20 age). Mid-parental height, target height attainment, and height velocity were evaluated. Statistical analysis used t-tests and chi-square tests (p <0.05).

RESULTS

Of the 248 children with DS, 63.7% had hypothyroidism, predominantly subclinical (85.4%). No cases of acquired or secondary hypothyroidism were found. Children with DS had significantly lower mean height z-scores (−1.91 ± 1.45 vs. −0.54 ± 1.25; p <0.001), borderline lower height velocity (5.80 ± 2.15 vs. 6.92 ± 3.56 cm/year; p = 0.050), and fewer achieved target height (35% vs. 64.1%; p = 0.030) compared to controls. Baseline characteristics were similar between groups. Thyroid ultrasound showed normal anatomy in 50%, hypoplasia in 28.6%, and nodules in 21.4%.

Common comorbidities included congenital heart disease (78.4%), pulmonary complications (16.2%), and other anomalies (32.4%).

CONCLUSION

Subclinical hypothyroidism was the predominant subtype in DS. Affected children demonstrated poorer growth, with reduced height z-scores, slower growth velocity, and lower likelihood of achieving target height.

EP_P006

CLINICAL CHARACTERISTICS AND OUTCOMES OF PAEDIATRIC HYPERTHYROIDISM: A 10-YEAR SINGLE CENTRE RETROSPECTIVE COHORT STUDY FROM NEGERI SEMBILAN

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INTRODUCTION

The commonest cause of paediatric hyperthyroidism is Graves' disease. Previous studies in the Malaysian context haven't captured data from Negeri Sembilan. This study aims to describe the demographic and clinical features of children with hyperthyroidism from Hospital Tuanku Jaafar.

METHODOLOGY

A retrospective cohort study collating socio-demographic and clinical data from electronic medical records was conducted. Statistical analysis was conducted using Microsoft Excel (v2602).

RESULTS

Fourteen children between 2015 and 2025 were identified. Females comprised 93% (n = 13). The mean age at diagnosis was 9.76 ± 3.4 years, with 64% (n = 9) aged >10 years. Malay ethnicity comprised 64% (n = 9). Family history was positive in 43% (n = 6). Three (21%) children were detected