

Paediatric E-Poster

EP_P001

EVALUATION OF ACANTHOSIS NIGRICANS AS A PREDICTIVE CLINICAL MARKER FOR METABOLIC RISK IN CHILDREN WITH OBESITY

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INTRODUCTION

Childhood obesity is associated with significant metabolic morbidity, particularly insulin resistance (IR) and metabolic syndrome. Acanthosis nigricans (AN) is frequently observed in children with obesity and may represent a practical clinical marker of IR. This study evaluated the association between AN, IR, and metabolic complications in paediatric obesity.

METHODOLOGY

A retrospective review was performed of 148 children (88 males, 60 females; median age 12 years) attending the Paediatric Obesity Clinic at Universiti Malaya Medical Centre. Data included anthropometry, AN grading, family history, HOMA-IR, and metabolic screening. Outcomes assessed were dyslipidemia, metabolic-associated fatty liver disease (MAFLD), hypertension, obstructive sleep apnea syndrome (OSAS), and glucose dysregulation. Comparisons were made between children with and without AN at baseline and over follow-up (median 2.4 years).

RESULTS

AN was present in 81.8% of patients, and 82.4% had central obesity. Increasing AN grade was significantly associated with higher BMI SDS and American Academy of Paediatrics obesity class ($p < 0.05$). AN was more prevalent among Malay and Indian children and was associated with a family history of obesity ($p < 0.05$). Baseline metabolic abnormalities were common, including dyslipidemia (high triglycerides 31.8%, low HDL 30.4%, high LDL 29.7%), MAFLD (23.6%), OSAS (31.1%), and glucose dysregulation (27.7%), with no significant difference between groups. During follow-up, children with AN and more severe obesity developed significantly more metabolic complications, particularly MAFLD and OSAS, with increased requirement for non-invasive ventilation ($p < 0.05$). Higher HOMA-IR was associated with glucose dysregulation but was not independently associated with AN.

CONCLUSION

AN is strongly associated with greater adiposity and predicts the progression of metabolic complications in children with obesity. Routine assessment of AN may help

identify high-risk patients who would benefit from early, intensive intervention to reduce long-term cardiometabolic morbidity.

EP_P002

PHENOTYPIC SPECTRUM AND GONADAL OUTCOMES IN CHILDREN WITH 45,X/46,XY MOSAICISM: A LONGITUDINAL COHORT STUDY

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INTRODUCTION

45,X/46,XY mosaicism is a rare disorder of sex development characterized by marked phenotypic variability, ranging from typical male or female genitalia to significant genital ambiguity. Data describing long-term clinical outcomes in affected patients remain limited. This study aimed to characterize the phenotypic spectrum and longitudinal outcomes of patients with 45,X/46,XY mosaicism, focusing on growth, associated comorbidities, and gonadal pathology.

METHODOLOGY

We conducted a retrospective longitudinal study at a tertiary paediatric endocrine referral centre between January 2006 and January 2025. Patients with cytogenetically confirmed 45,X/46,XY mosaicism or related variants were included. Clinical presentation, karyotype, anthropometry, hormonal profiles, imaging findings, and gonadal histology were reviewed. External genital phenotype was quantified using the External Masculinization Score (EMS). Height was expressed as standard deviation scores (SDS) relative to population norms and compared with genetic potential.

RESULTS

Fourteen patients (10 males, 4 females) with 45,X/46,XY mosaicism were identified. Ambiguous genitalia was the predominant presenting feature, observed in 11 of 12 patients with available phenotype data. Growth impairment was common; median height SDS was -2.62 (range -3.26 to -2.00) in females and -0.46 (range 0.13 to -1.70) in males, both below expected genetic potential. Associated congenital anomalies were present in 25% of patients, including cardiac and renal abnormalities. Male patients demonstrated variable degrees of undervirilization with a median EMS of 4.5 (3.75-7.25). Müllerian duct remnants were identified in 66% of patients on imaging. Testicular microlithiasis was observed in two males. During follow-up, two patients underwent gender reassignment. Histological analysis of 14 gonads obtained via gonadectomy or biopsy

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 multi-disciplinary 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