EP_P001

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

Annie Leong[1], Nurshadia Samingan[1], Muhammad Yazid Jalaludin[1], Azriyanti Anuar Zaini[1]
[1]Paediatric Department, University Malaya Medical Centre

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

Vaidevi Poospalinggam[1], Janet Hong[1], Arini Nuran[1], Nalini M Selveindran[1]
[1]Paediatric Endocrine Unit, Hospital Putrajaya

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