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CLINICAL DIVERSITY AND GENETIC FINDINGS IN CHILDREN WITH DIFFERENCE OF SEX DEVELOPMENT

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INTRODUCTION
Difference of sex development (DSD) is a rare disease. Clinical classification is difficult due to similar phenotypes with different genetic etiologies. The study aimed to determine the clinical diversity and genetic diagnosis of patients with DSD in Hospital Tunku Azizah (HTA), Kuala Lumpur.

METHODOLOGY
Children with DSD (except Turner syndrome and congenital adrenal hyperplasia) who attended the Paediatric Endocrine Clinic, HTA between January to December 2021 were included. Data including karyotypes and whole exome sequencing results were retrospectively reviewed.

RESULTS
Twenty-seven children were identified: 23 with 46,XY DSD, two 46,XX DSD and two sex chromosome DSD. Majority (59.3%) presented at the neonatal period; the rest during prepubertal (33.3%) and pubertal (7.4%) ages. All 16 neonates presented with ambiguous genitalia, with external genitalia score (EGS) 7.9 ±1.6. All were assigned as male, and 93.8% (15/16) were 46,XY. Nine children presented at the prepubertal period, with mean age of 4.8 ± 3.9 years. Of these, 55.6% (5/9) were brought up as male with EGS 5.7 ± 2.9. Six were 46,XY, two 45,XO/46,XY and one 46,XX. The reasons for referral were middle/proximal hypospadias (50%), cryptorchidism (37.5%), micropenis (25%) and virilisation (12.5%). Two adolescents (46,XY) presented at puberty with mean age of 11.5 ± 0.7 years and EGS 5.8 ± 3.9. The most common diagnosis was gonadal dysgenesis (10/27, 37%), followed by androgen insensitivity syndrome (AIS) (33.3%), 5-alpha reductase (5αR) deficiency (11.1%), and ovotesticular (11.1%) and mixed gonadal dysgenesis (MGD) (7.4%). Nineteen children (70.4%) had genetic testing. Two were found to have MGD (45,XO/46,XY), two with gonadal dysgenesis (WT1 gene), one with 5αR deficiency (NR5A1) and one with AIS (AR).

CONCLUSION
We observed a wide spectrum of DSD in our clinical setting. An accurate genetic diagnosis is crucial to predict long term outcomes. Reanalysis maybe required in the future for unsolved cases.