ABSTRACT

Background: Differences of sex development (DSD) are heterogeneous congenital conditions involving limited molecular testing increases the importance of clinical, cytogenetic, hormonal, and radiological data.
Objective: To describe and integrate the clinical, genetic, hormonal, and radiological profiles of individuals with DSD
Method: This descriptive cross-sectional study reviewed records from CEBIOR, Diponegoro University, from 2020 to March 2026. Individuals aged 0–18 years with ambiguous genitalia and/or delayed testosterone, FSH, LH, estradiol, ultrasonography descriptively.
Result: Among 145 probands, 133 (91.72%) had a 46,XY karyotype and 12 (8.28%) had 46,XX. Median age at admission was 1.33 years; 86.89% were reared as male and 83.45 ,XX probands. Presumptive diagnoses were non-specific 46,XY DSD (75.9%), gonadal dysgenesis (11.0%), syndromic DSD (6.2%), AIS (3.4%). WES identified the likely pathogenic proband with AIS.
Conclusion: DSD in this cohort showed substantial clinical, genetic, hormonal, and radiological heterogeneity. Integration of these profiles enabled presumptive diagnostic aetiological and provide more accurate genetic counseling regarding inheritance, recurrence risk, and familial implications.

Keywords: differences of sex development; karyotype; hormonal profile; ultrasonography; presumptive diagnosis.