

DIAGNOSTIC PERFORMANCE OF THE EXTERNAL GENITALIA SCORE FOR SEX

ASSIGNMENT CONSIDERATION IN CHILDREN WITH ATYPICAL GENITALIA:

A PRELIMINARY STUDY AT DR. SARDJITO HOSPITAL

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Abstract

Background: Disorders of Sex Development (DSD) may present with atypical genitalia. Karyotyping is the gold standard for the determination of chromosomal sex, but it may not be readily available. The External Genitalia Score (EGS) assesses genital masculinization. This study evaluated EGS for distinguishing 46,XY from 46,XX karyotypes in children with atypical genitalia.

Methods: A cross-sectional study was conducted at Dr. Sardjito General Hospital, Indonesia. The EGS of children with atypical genitalia were assessed by two investigators before the karyotypes as the reference standard were performed. The cut-off for EGS was determined using receiver operating characteristic curve (ROC). The cut-off was used to assess sensitivity, specificity, positive predictive value, negative predictive value, and accuracy

Results: Eighteen children were included: 7 with 46,XX and 11 with 46,XY karyotypes. Inter-rater agreement between the two investigators was Cohen's kappa (κ) = 0.682, $p < 0.001$. Median total EGS of 46,XX children was lower than 46,XY children (1.0 [IQR 0.0–4.0] vs. 7.5 [IQR 4.0–12.0], $p < 0.001$). Total EGS showed discriminative ability (AUC 0.994; 95% CI 0.960–1.000, $p < 0.001$). A cut-off of 4.5 yielded 90.9% sensitivity, 100% specificity, 100% positive predictive value, 87.5% negative predictive value, and 94.4% accuracy.

Conclusion: EGS demonstrated good discriminative ability for distinguishing chromosomal sex in children with atypical genitalia and may support early clinical assessment. Further studies with larger and more diverse karyotypes are required to confirm these findings and validate the proposed cut-off value.

Keywords: *Disorders of Sex Development, Atypical Genitalia, External Genitalia Score, Karyotype*

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INTISARI

Latar Belakang: *Disorders of Sex Development* (DSD) dapat bermanifestasi sebagai *atypical genitalia*. Kariotipe merupakan standar baku untuk menentukan jenis kelamin kromosom, tetapi pemeriksaan ini tidak selalu tersedia. *External Genitalia Score* (EGS) menilai derajat maskulinisasi genitalia. Penelitian ini mengevaluasi kemampuan EGS dalam membedakan *karyotype* 46,XY dan 46,XX pada anak dengan *atypical genitalia*.

Metode: Penelitian potong lintang dilakukan di RS Dr. Sardjito, Indonesia. EGS pada anak dengan genitalia atipikal dinilai oleh dua peneliti sebelum pemeriksaan *karyotype* sebagai standar rujukan. Nilai *cut-off* EGS ditentukan menggunakan kurva *receiver operating characteristic* (ROC), kemudian digunakan untuk menilai sensitivitas, spesifisitas, nilai prediksi positif, nilai prediksi negatif, dan akurasi.

Hasil: Sebanyak 18 anak diikutsertakan, terdiri atas 7 anak dengan *karyotype* 46,XX dan 11 anak dengan 46,XY. Kesepakatan antarpemeriksa menunjukkan Cohen's kappa (κ) = 0,682; $p < 0,001$. Median total EGS pada kelompok 46,XX lebih rendah dibandingkan 46,XY (1,0 [IQR 0,0–4,0] vs. 7,5 [IQR 4,0–12,0]; $p < 0,001$). EGS menunjukkan kemampuan diskriminatif yang sangat baik (AUC 0,994; IK95% 0,960–1,000; $p < 0,001$). Nilai *cut-off* 4,5 menghasilkan sensitivitas 90,9%, spesifisitas 100%, nilai prediksi positif 100%, nilai prediksi negatif 87,5%, dan akurasi 94,4%.

Kesimpulan: EGS memiliki kemampuan diskriminatif yang baik dalam membedakan jenis kelamin kromosom pada anak dengan *atypical genitalia* dan dapat mendukung penilaian klinis awal. Penelitian lebih lanjut dengan jumlah sampel dan variasi kariotipe yang lebih besar diperlukan untuk mengonfirmasi temuan serta memvalidasi nilai *cut-off* tersebut.

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