Hubungan antara tampilan genitalia eksterna dengan jenis kromosom seks pada pasien amenore primer di RS Cipto Mangunkusumo, Jakarta = Relationship between external genitalia display and sex chromosome types in primary amenorrhea patients at Cipto Mangunkusumo Hospital, Jakarta

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Abstrak

Latar belakang: Amenore primer merupakan gangguan pada siklus menstruasi wanita. 11,1% wanita remaja menunjukkan keluhan ginekologi terkait menstruasi. Prevalensi kasus amenore primer terjadi pada 0,1â2% wanita usia reproduksi.
Amenore primer dapat disebabkan sex reversal female (kromosom 46XY) atau
sindrom Turner (45X). Abnormalitas perkembangan genitalia eksterna merupakan
manifestasi dari gangguan kromosom. Belum ada penelitian yang mengkaji hubungan antara presentasi genitalia eksterna dengan genotip kromosom seks pada pasien amenore primer di Rumah Sakit Cipto Mangunkusumo (RSCM), Jakarta.
Tujuan: Mengetahui tampilan genitalia eksterna pada pasien amenore primer di RSCM, Jakarta dan hubungannya dengan kejadian ambiguitas genitalia. Metode: Penelitian ini dilakukan desain studi dengan potong lintang secara retrospektif menggunakan rekam medis pasien amenore primer Departemen Obstetri dan Ginekologi RSCM periode Januari tahun 2018-2020. Data diolah
menggunakan program Statistical Package for the Social Sciences (SPSS) versi 20.
Uji hipotesis akan dilakukan menggunakan uji Chi-Square atau Fisherâs exact test.
Hasil: 65 rekam medis amenore primer yang melakukan analisa genetika berhasil dikumpulkan. 90,77% (n=59) kasus amenore primer bergenitalia eksterna normal dan 9,23% (n=9) bergenitalia ambigu. Seluruh kasus genitalia ambigu
berkarakteristik pembesaran klitoris dengan 1 kasus juga diamati phallus genital. Hanya 53,8% kasus yang memiliki kromosom perempuan normal, 46XX. Hubungan signifikan didapatkan antara ambiguitas genitalia eksterna dan
abnormalitas kromosom seks (p=0,042) Simpulan: Terdapat hubungan yang signifikan antara ambiguitas genitalia eksterna serta tidak normalnya hasil analisa kromosom seks pada pasien amenore primer di Departemen Obstetri dan Ginekologi RSCM periode Januari tahun 2018-2020.
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Background: Primary amenorrhea is a disorder of a woman's menstrual cycle. 11.1% of adolescent women showed gynecological complaints related to
menstruation. The prevalence of primary amenorrhea cases occurs in 0.1â2% of women of reproductive age. Primary amenorrhea can be caused by female sex reversal (chromosome 46XY) or Turner syndrome (45X). Abnormalities in the development of external genitalia are a manifestation of chromosomal disorders. There are no studies that have examined the relationship between external genitalia presentation and sex chromosome genotypes in primary amenorrhea patients at Cipto Mangunkusumo Hospital (RSCM), Jakarta. Goal: Finding the characteristic the appearance of external genitalia in primary amenorrhea patients at

RSCM, Jakarta and its relationship with the incidence of genital ambiguity Method: This study was conducted with a retrospective cross-sectional study design using the medical records of primary amenorrhea patients from the Department of Obstetrics and Gynecology RSCM January 2018-2020. Data were processed using the Statistical Package for the Social Sciences (SPSS) program version 20. Hypothesis testing will be carried out using the Chi-Square test or Fisher's exact test. Result: 65 medical records of primary amenorrhea that performed genetic analysis were collected. 90.77% (n = 59) cases of primary amenorrhea with external genitalia were normal and 9.23% (n = 9) had ambiguous genitalia. All cases of ambiguous genitalia were characterized by clitoral enlargement with 1 case also observed for the genital phallus. Only 53.8% of cases had a normal female chromosome, 46XX. There was a significant relationship between external genital ambiguity and sex chromosome abnormalities (p = 0.042). Conclusion: There is a significant relationship between external genital ambiguity and abnormal results of sex chromosome analysis in primary amenorrhea patients in the Obstetrics and Gynecology Department of RSCM for the period January 2018-2020 (p<0,05).