Journal of Andrology, Vol. 24, No. 1, January/February 2003

Clinical and Diagnostic Features of Patients With Suspected Klinefelter Syndrome

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Klinefelter syndrome, with an incidence of 1:600 male newborns, is the most frequent form of male hypogonadism. However, despite its relatively high frequency, the syndrome is often overlooked. To prevent such oversights, the clinical features should be better characterized, and simple screening tests should be used more frequently. In a cohort of 309 patients suspected of having Klinefelter syndrome, we evaluated the clinical symptoms as well as the diagnostic value of the Barr body test for screening procedures. On the basis of chromosome analysis, 85 patients (group I) were diagnosed as having Klinefelter syndrome, and 224 patients had a 46,XY karyotype (group II). Barr body analysis revealed a specificity of 95% and a sensitivity of 82% for the diagnosis of Klinefelter syndrome. General features (eg, reason for admission, age, age of the parents, body weight, and frequency of maldescended testes) were not different between the groups, except that group I had a higher proportion of patients with a lower educational background. Compared to group II, patients with Klinefelter syndrome were taller ($P < .001$); had smaller testis volumes ($P < .0001$), higher follicle-stimulating hormone (FSH) and luteinizing hormone (LH) values; and carried a tendency for less androgenic phenotype and secondary hair distribution. Testosterone, estradiol, sex hormone—binding globulin (SHBG), and prostate-specific antigen (PSA) serum levels as well as prostate volume were not significantly different between the groups. In patients who provided an ejaculate, azoospermia was found in 54% of the patients in group II and in 93% of the patients with Klinefelter syndrome. Although not exclusively characteristic for Klinefelter syndrome, the combination of low testicular volume and azoospermia, together with elevated gonadotropins, is highly indicative for a Klinefelter syndrome and should stimulate further clinical investigations. Barr body analysis provides a quick and reliable screening test, which, however, must be confirmed by karyotyping.

Key words: Male hypogonadism, testosterone substitution, 47,XXY, Barr body analysis

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