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Size of the exon 1-CAG repeats of the androgen receptor gene employed as a molecular marker in the diagnosis of Turner syndrome in girls with short stature

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ABSTRACT. Turner syndrome (TS) is one of the most common chromosomal abnormalities among girls. Complete monosomy of X chromosome is responsible for almost 50% of all cases of TS, and mosaicism and X anomaly are detected in the other half. It has already been demonstrated that early diagnosis of these children allows appropriate growth hormone treatment with better final height prognosis and introduction of estrogen at an ideal chronological age. Sixty-four short-stature girls were selected and the clinical data obtained were birth weight and height, weight and height at the first medical visit and target height. Other clinical data including cardiac and renal abnormalities, otitis, Hashimoto thyroiditis, cubitus valgus, short neck, widely separated nipples, and pigmented nevi were obtained from the patients' medical records. The aim of the present study was to evaluate the screening of a group of short-stature girls for TS based on the number of CAG repeats of the androgen receptor gene analyzed by

GeneScan software. Patient samples with two alleles (heterozygous) were 49/64 (76.5%) and with one allele (homozygous) were 15/64 (23.5%). A karyotype was determined in 30 patients, 9 homozygous and 21 heterozygous. In the homozygous group, 6/9 were 45,X and 3/9 were 46,XX. In the heterozygous group, 17/21 were 46,XX, and 4/21 were TS patients with mosaicism (45,X/46,XX; 45,X/46XiXq; 46XdelXp). The pattern obtained by GeneScan in two patients with mosaicism in the karyotype was an imbalance between the peak heights of the two alleles, suggesting that this imbalance could be present when there is a mosaicism. The frequency of TS abnormalities (18.7%) did not differ between TS and 46,XX girls. Thus, it is important to accurately assess the incidence of TS in growth-retarded girls, even in the absence of other dysmorphisms. In this study, we diagnosed 6 cases of TS 45,X (9.4%) by molecular analysis, with a 100% sensitivity and 85% specificity. This molecular analysis was able to detect all cases of TS 45,X and the majority of mosaicisms, without the need for more X chromosome markers. In conclusion, determining the number of CAG repeats of the androgen receptor gene analyzed by GeneScan was a fast method with high sensitivity for the detection of TS 45,X, suggesting that it could be interesting as a method for screening a population of growth-retarded girls.

Key words: Turner syndrome; Androgen receptor; Molecular diagnosis; Short stature

