



Utility of STR markers for the molecular diagnosis of a large Brazilian family with Charcot-Marie-Tooth disease

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Genet. Mol. Res. 7 (4): 1179-1185 (2008)

Received July 28, 2008

Accepted August 21, 2008

Published October 28, 2008

ABSTRACT. Charcot-Marie-Tooth type 1A disease (CMT1A) is most frequently caused by a tandem DNA duplication of a 1.4-Mb genomic fragment in the 17p11.2-12 chromosomal region. The disease is probably the product of a dosage effect of the peripheral myelin protein 22 gene located within the duplicated segment. We sought to study the largest reported Brazilian family with suspected diagnosis of CMT1A using eight short tandem repeat microsatellite markers. In addition, we analyzed the informativeness of these markers in the normal Brazilian population. The duplication was found in 12 members of the family. In two patients with CMT1A symptoms, the duplication was not detected, and one asymptomatic subject showed the duplication. D17S2230, D17S9B, D17S2220, D17S2227, D17S9A, and D17S4A markers showed the highest heterozygosity rates, and D17S2228 and

D17S2224 markers were the least informative in our analysis.

Key words: Charcot-Marie-Tooth 1A; 17p11.2-p12 duplication;
Short tandem repeat markers; Molecular diagnosis; Brazilian population