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A985G mutation incidence in the medium-chain acyl-CoA dehydrogenase (MCAD) gene in Brazil

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ABSTRACT. In view of the serious consequences of medium-chain acyl-CoA dehydrogenase (MCAD) deficiency and the absence of information about its incidence in the Brazilian population, we examined the frequency of the A985G mutation in the MCAD gene. A retrospective analysis was made of data on 1722 individuals (844 females) genotyped for the A985G mutation in the MCAD gene, using genomic DNA extracted from peripheral blood leukocytes and genotyping with PCR-RFLP; 0.41% of these individuals were heterozygous for the A985G mutation. The mutant homozygous genotype was not found. The 985G mu-

tant and 985A normal alleles had allelic frequencies of 0.0020 and 0.9980, respectively. Given the A985G allele frequency, genotyping would be recommended in cases of family history of MCAD deficiency and sudden infant death syndrome, and when there is suspicion of medium-chain fatty acid metabolic alterations; genetic counseling should be offered in cases involving 985GG and A985G individuals and consanguineous marriages.

Key words: A985G mutation; MCAD deficiency; Molecular screening; Medium-chain acyl-CoA dehydrogenase deficiency; Brazil

