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Variations in genotype-phenotype correlations in phenylketonuria patients

L.L. Santos[1,4], C.G. Fonseca[1], A.L.P. Starling[3], J.N. Januário[3], M.J.B. Aguiar[3], M.G.C.D. Peixoto[2] and M.R.S. Carvalho[1]

[1]Departamento de Biologia Geral, Instituto de Ciências Biológicas, Universidade Federal de Minas Gerais, Belo Horizonte, MG, Brasil
[2]Embrapa Gado de Leite, Juiz de Fora, MG, Brasil
[3]NUPAD - Núcleo de Ações e Pesquisa em Apoio Diagnóstico, Faculdade de Medicina, Universidade Federal de Minas Gerais, Belo Horizonte, MG, Brasil
[4]Universidade Federal de São João Del-Rei, Divinópolis, MG, Brasil

Corresponding author: M.R.S. Carvalho
E-mail: mraquel@icb.ufmg.br

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ABSTRACT. Phenylalanine hydroxylase deficiency is a trait inherited in an autosomal recessive pattern; the associated phenotype varies considerably. This variation is mainly due to the considerable allelic heterogeneity in the phenylalanine hydroxylase enzyme locus. We examined the genotype-phenotype correlation in 54 phenylketonuria (PKU) patients from Minas Gerais, Brazil. Two systems were used. The first was a phenotype prediction system based on arbitrary values (AV) attributed to each mutation and the second was a correlation analysis. An AV was assigned to each mutation: AV = 1 for classical PKU mutation; AV = 2 for moderate PKU mutation; AV = 4 for mild PKU mutation, and AV = 8 for non-PKU hyperphenylalaninemia mutation. The observed phenotype for AV analysis was the clinical diagnosis established by the overloading phenylalanine test. Among the 51 PKU patients that we analyzed based on this trait, in 51% the predicted phenotype did not match the observed phenotype; the highest degree of concordance was found in patients with null/null genotypes. The genotype was observed to be a good predictor of the clinical course of the patients and significant correlations were

found between phenylalanine values at first interview and predicted residual activity, genotype and arbitrary value sum.

Key words: Phenylketonuria; Minas Gerais, Brazil; Genotype; Phenotype; Correlation

