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High Frequency of Severe Phenylketonuria in Jalisco, Mexico

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ABSTRACT This study reports the epidemiological findings collected during 11.5 years of the genotypes, metabolic and clinical phenotypes, of phenylketonuria (PKU) in twenty-two Mexican children in the state of Jalisco. The phenylalanine hydroxylase (*PAH)* variants were identified in 17/22 PKU cases. Four cases had mild hyper-phenylalanine (MHPA), two had mild PKU, one subject had moderate PKU and ten cases had classic PKU. Twelve variants of the *PAH* gene were identified: c.60+5G>T with 47.1 percent followed by c.441+5G>T, c.508C>G and c.1241A>G with 8.8 percent each; c.106611G>A with 5.9 percent and other variants with 2.9 percent each. A new pathogenic missense mutation is reported in c.791A>G. The researchers' study suggests that the population of Jalisco has a spectrum not found in the rest of the country with a genetic heterogeneity that has shown more severe variants.