

## Allelic frequency of DPYD genetic variants: implementation of a genotyping test in Mexican population

Diana Cristina Pérez-Ibave  
*Universidad Autónoma de Nuevo León*

Carlos H. Burciaga-Flores  
*Universidad Autonoma de Nuevo Leon*

Valeria Jimena Gómez-Ordaz  
*Universidad Autonoma de Nuevo Leon*

Noé Israel Oliva-García  
*Universidad Autónoma de Nuevo León*

Vanessa N. Ortiz-Murillo  
*Universidad Autonoma de Nuevo Leon*

*See next page for additional authors*

Follow this and additional works at: <https://scholarworks.utrgv.edu/somrs>



Part of the [Medicine and Health Sciences Commons](#)

---

### Recommended Citation

Pérez-Ibave, Diana Cristina; Burciaga-Flores, Carlos H.; Gómez-Ordaz, Valeria Jimena; Oliva-García, Noé Israel; Ortiz-Murillo, Vanessa N.; Alvarado Villarreal, Francisco; Cortes-Alfaro, Jonatán Isaí; Burguete-Torres, Alan; Alcorta-Núñez, Fernando; Vidal Gutiérrez, Oscar; and Garza-Rodríguez, María Lourdes, "Allelic frequency of DPYD genetic variants: implementation of a genotyping test in Mexican population" (2024). *Research Symposium*. 102.

<https://scholarworks.utrgv.edu/somrs/2023/posters/102>

This Poster is brought to you for free and open access by ScholarWorks @ UTRGV. It has been accepted for inclusion in Research Symposium by an authorized administrator of ScholarWorks @ UTRGV. For more information, please contact [justin.white@utrgv.edu](mailto:justin.white@utrgv.edu), [william.flores01@utrgv.edu](mailto:william.flores01@utrgv.edu).

---

**Presenter Information (List ALL Authors)**

Diana Cristina Pérez-Ibave, Carlos H. Burciaga-Flores, Valeria Jimena Gómez-Ordaz, Noé Israel Oliva-García, Vanessa N. Ortiz-Murillo, Francisco Alvarado Villarreal, Jonatán Isai Cortes-Alfaro, Alan Burguete-Torres, Fernando Alcorta-Núñez, Oscar Vidal Gutiérrez, and María Lourdes Garza-Rodríguez

## Allelic frequency of *DPYD* genetic variants: implementation of a genotyping test in Mexican population

Diana Cristina Pérez- Ibave<sup>1</sup>, Carlos Horacio Burciaga-Flores<sup>1</sup>, Valeria Jimena Gómez-Ordaz<sup>1</sup>, Noé Israel Oliva-García<sup>1</sup>, Vanessa Natalí Ortiz-Murillo<sup>1</sup>, Francisco Alvarado Villarreal<sup>1</sup>, Jonatán Isai Cortes-Alfaro<sup>1</sup>, Alan Burguete-Torres<sup>1</sup>, Fernando Alcorta-Nuñez<sup>1</sup>, Oscar Vidal Gutiérrez<sup>1</sup>, and María Lourdes Garza-Rodríguez<sup>1\*</sup>.

1. Servicio de Oncología, Centro Universitario Contra el Cáncer (CUCC), Hospital Universitario "Dr. José Eleuterio González", Universidad Autónoma de Nuevo León, Monterrey, Nuevo León, 66451 México.

**Background:** Fluoropyrimidine-based (FP) chemotherapy is extensively used to treat solid cancers, including colorectal and breast cancer. A dihydropyrimidine dehydrogenase (DPD) enzyme deficiency, encoded by the dihydropyrimidine dehydrogenase (*DPYD*) gene, increases the risk of severe toxicity. FP toxicity affects about 30-40% of patients, which in some cases may be lethal. FPs have been used for over 50 years, and an estimated 2 million cancer patients are treated with FP drugs annually. In particular, FPs remain among the most effective drugs for treating GI malignancies, including colorectal cancer (CRC) (1.8 million), gastric (1 million), and pancreatic cancer (n=460,000). In Mexican oncology practice, FP and capecitabine chemotherapies are the most common drugs for gastrointestinal, head and neck, and breast tumors. *DPYD* genotyping aims to identify variants that lead to DPD deficiency. We implemented a seven-allelic genotyping test and analyzed the frequency in the Mexican population.

**Methods:** We included seven *DPYD* variants: c.1129-5923C->G, c.2846A->T associated with increased risk toxicity (reduced activity), and c.1156G->T, c.1905+1G->A, c.1679T->G, c.1898delC, and c.299\_302delTCAT associated with high risk for FP toxicity (no activity or significantly reduced activity). Genomic DNA was isolated from 280 subjects: 36 cancer patients and 244 non-cancer subjects. We analyzed *DPYD* variants by real-time PCR (c.1156G->T, c.2846A->T, and c.1129-5923C->G) and Sanger sequencing (c.1905+1G->A, c.1679T->G, c.1898delC and c.299\_302delTCAT) The allele frequency was calculated for each variant.

**Results:** For Sanger sequencing, primers were designed to amplify four variants. Amplified products of the expected size were obtained. Three variants were amplified using TaqMan probes and synthetic positive controls for both alleles. We found the c.1129-5923C>G variant in the heterozygous state in 1% (n= 3), and the c.2846A>T variant was found in 0.33% (n=1) of the participants. We did not found the rest of the variants in the Mexican population.

**Conclusions:** The allele frequency for two of the seven analyzed variants (c.1129-5923C->G and the c.2846A>T) was higher than reported for the global population (0.00476, and 0.005166). *DPYD* genotyping may help identify patients at higher risk of developing severe FP toxicity. Personalized medicine allows oncologists to modify the treatment before it begins.