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PGMD: A comprehensive pharmacogenomic database for personalized medicine and drug discovery

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The Pharmacogenomic Mutation Database (PGMD) is a comprehensive manually curated pharmacogenomics database. The aim of this database is to provide a comprehensive resource for all variants that have been reported to have a pharmacogenomic effect in human studies and to describe those variants by exact genomic location and sequence alterations for application to NGS data analysis. The database is designed to contain extensive information as evidence for these associations including provenance of every observation. Two major sources of PGMD data are peer reviewed literature and FDA drug labels. PGMD curators capture information on exact genomic location and sequence changes resulting phenotype, drugs administered, patient population, study design, disease context, statistical significance and other properties of reported pharmacogenomic variants. Variants are annotated into functional categories basing on their influence on pharmacokinetics, pharmacodynamics, efficacy or clinical outcome. The current release of PGMD includes nearly 140000 unique pharmacogenomic observations, covering all 24 disease super classes and 1377 drugs. Over 2800 genes have associated pharmacogenomic variants including genes in proximity to intergenic variants. PGMD is optimized for use in annotating next generation sequencing data by providing genomic coordinates for all covered variants including SNPs, insertions, deletions, haplotypes, diplotypes, VNTRs, copy number variations and structural variations.

Biography

Alexander Kaplun has completed his PhD from Ben Gurion University and Postdoctoral studies from Karmanos Cancer Institute. He is a Senior Scientist, Advanced Genomics Integrated Solutions at QIAGEN Bioinformatics, Global Leader in development and distribution of clinical and biological software tools and databases. He has published more than 20 papers in peer-reviewed journals.

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