Pharmacogenetics in Routine Clinical Diagnostics: Updated Experience from the Central Laboratory of the University Hospital of Innsbruck

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Introduction. Pharmacogenetic genotypes play a major role in interindividual variability in drug response, including differences in absorption, metabolism and pharmacodynamics. Consequently, pharmacogenetic testing enables the optimization of drug efficacy while minimizing drug-related toxicity. Although awareness of pharmacogenetics is increasing, its routine application in daily clinical practice has not yet been widely established.
Methods. Since 2012, several pharmacogenetic parameters have been analyzed in the Central Laboratory of the University Hospital of Innsbruck using PCR-based methods. These include *thiopurine methyltransferase* (TPMT), *dihydropyrimidine dehydrogenase* (DPD), *cytochrome P450* (CYP) enzymes CYP2C19, CYP2C9, CYP3A4/5, and CYP2D6, *UDP-glucuronosyltransferase 1A1* (UGT1A1), and *solute carrier organic anion transporter family member 1B1* (SLCO1B1).
Results. In 2025, the most frequently requested pharmacogenetic parameters were DPD (417 tests, 52%) and TPMT (296 tests, 37%), followed by CYP enzymes (72 tests, 9%), UGT1A1 (14 tests, 2%), and SLCO1B1 (6 tests, 1%). While the number of requests for TPMT, CYP2C9, CYP3A4/5, and SLCO1B1 remained largely stable over the past seven years, marked increases were observed for several parameters. Requests for DPD testing increased substantially from 2019 onward, following guideline recommendations for DPD genotyping prior to fluoropyrimidine dosing. A significant rise in CYP2C19 testing was noted beginning in 2023, coinciding with the approval of mavacamten in the European Union. Similarly, UGT1A1 testing increased from 2022 onward due to growing scientific and clinical awareness of the importance of pharmacogenetic testing prior to treatment with sacituzumab, govitecan and irinotecan.
Conclusion. Pharmacogenetic testing has been successfully implemented in our laboratory. A marked increase in selected tests, particularly DPD, reflects the growing clinical relevance of pharmacogenetics driven by clinical guidelines, regulatory approvals, and therapeutic advances, underscoring its increasing importance in personalized medicine. Although awareness of pharmacogenetic testing is rising, there remains substantial potential for further improvement, and continued education of medical staff is essential.