

# Pharmacogenetics and Pharmacogenomics

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## 1 INTRODUCTION: PHARMACOGENETICS AND PHARMACOGENOMICS

The fields of pharmacogenetics, genomics, and drug transporters have profoundly impacted drug metabolism research by providing plausible mechanisms for interindividual variability in drug response and metabolism-related toxicity. They have provided tools with which to understand enzyme regulation, identify factors that affect drug exposure, the potential for drug–drug interactions, and species differences in drug disposition. Knowledge from these fields is being used to form the scientific basis for designing appropriate clinical studies and data interpretation, leading to the development and use of safer and more efficacious drugs.

Pharmacogenetics is the study of the effects of genetic differences between individuals on interindividual responses to medicines (Ginsburg *et al.*, 2005; Eichelbaum, Ingelman-Sundberg and Evans, 2006). It is an old discipline, which has been invigorated by an increased understanding of molecular biology and development of associated technological tools with which to study these mechanisms (Goldstein, Tate and Sisodiya, 2003). In its simplest form, genetics is the scientific study of heredity, dating back to Mendel, who showed that the inheritance pattern of certain traits in pea plants followed simple statistical rules and described a fundamental unit of heredity, which he called allele. While Mendel used *allele* to refer to what we now know as gene, allele is currently used to denote a specific variant of a particular gene. Pharmacogenetics can therefore be viewed as a more specific and narrow subfield of genetics. Though an old discipline, the term having been around since the 1950s has enjoyed a new lease of life in the last 10–20 years, mainly due to advances in our understanding of the impact of genetics on efficacy and safety of drugs, emergence of global tools with which to study multiple genes at any given time, and the completion of the human genome project.

Pharmacogenomics is a more recent term, coined to define a more holistic or global approach, in which the expression levels, regulation, functions, and interactions of