

Interestingly, the role of biotechnology in public health and drug discovery and development is growing fast. 20% of the pharmaceutical products already marketed nowadays are of derived from biotech approaches, as well as the 40% of the molecules currently under registration and the 50% of drugs in phase of development and testing. Two of the main interesting fields for the application of biotechnology to the human health are pharmacogenetics/pharmacogenomics and research on orphan drugs.

As the name suggests, pharmacogenetics is the intersection of the fields of pharmacology and genetics. Simply stated, pharmacogenetics is the study of how genetic variations affect the ways in which people respond to drugs. These variations can manifest themselves as differences in the drug targets or as differences in the enzymes that metabolize drugs. A difference in the target will usually lead to differences in how well the drug works, whereas differences in metabolizing enzymes can result in differences in either efficacy or toxicity. It's also possible that genes not directly involved in a particular pathway could end up being predictive of clinical outcomes.

In 2000, completion of the human genome sequence was publicly applauded, although in reality it took a few more years to obtain the first full sequence data. Within 10 years, sequencing technology has advanced to an extent seemed unreachable at that time. Whilst, in 2000, it took many years to sequence just one human genome, today such sequencing can be done within 1-2 weeks. Human genetic variation has been catalogued in depth to at least 5% prevalence in Caucasians and efforts are underway to broaden this coverage to at least 1%. Haplotype maps have been constructed to extract the most informative SNPs for genetic studies. Genome-wide association studies based on 500,000 to 1 million such SNPs for mapping and gene detection efforts are the norm today. Genotype-guided therapy is emerging into mainstream medicine, setting the stage for a future of personalized medicine as compared to today's group-based medicine, where therapeutic efficacy is defined from mean effects in large-scale Phase III studies in humans.

Besides the potential of pharmacogenomics, another main topic of interest of the scientific community nowadays is represented by the molecule discovery in the field of orphan drugs. In Italy, 209 biotech projects are allowed by the Ministry of Health and 40 of them are focused on orphan drugs. An orphan drug is a pharmaceutical agent that has been developed specifically to treat a rare medical condition, the condition itself being referred to as an orphan disease. The assignment of orphan status to a disease and to any drugs developed to treat it is a matter of public policy in many countries, and has resulted in medical breakthroughs that may not have otherwise been achieved due to the economics of drug research and development.