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In some way, each person is like all others

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In some way unique, like no other.  
(Origin unknown)

All humans are equal, and yet so diverse. We share the same genetic information, with the exception of minute differences, yet we differ in our appearance, character, behavior, and health status. Our knowledge about the genetic bases of health status has rapidly changed over the last few years. The completion of the Human Genome Project provided us with a map of the 3 billion base pairs encoding the blueprint of human life. It is now possible to sequence the entire genome of an individual in a single day! However, while the DNA code of a single human may appear simple, the interpretation of this code for the purpose of understanding the intricacies of health and disease has proven to be tremendously complex. Advancements in molecular biology have determined that factors beyond the genetic code itself—for example, epigenetic regulation and noncoding RNAs—are integral to these processes. These discoveries, and the ever-expanding availability of clinical assays for genetic traits, have led to the emergence of the relatively new field of “molecular medicine” in which physicians and health care providers apply genetic information to maintain health, rapidly diagnose illness, and solve the problems associated with human disease.

All physicians in this age of molecular medicine must be well versed in the core principles of human genetics, which is the science of the mechanisms and principles through which genetic information determines health and disease. Searching for genetic changes and gene variants within a population, questioning the link between genotype and phenotype, probing the meaning of gene–gene and gene–environment interactions, investigating the role of somatic mutations in the formation of tumors, exploring the possibilities for prenatal diagnostics, and surveying the gene therapy for directed treatment strategies and preventative medicine are relevant for all fields of medicine. Human genetics serves as an important bridge between basic biology, on one hand, and practical clinical medicine, on the other. It is a “meta-discipline” that permeates all medical specialties. It helps with the diagnosis of a genetic predisposition to the development of a particular illness as well as the interdisciplinary care of affected individuals. It also serves to meet special needs for communicating information about the cause of an illness and its significance in the context of genetic counseling.

The 50 years that span the timeline of early genetic medicine, from the discovery of the DNA double helix by Watson and Crick (in 1953) to the publication of the sequence of 99.99% of the human genome (in 2003), can be thought of as the “pregenomic” period in medical history. As we approach the second decade of the “postgenomic” era, we have a greater appreciation of the complexities regulating the genomic sequence and the importance of understanding these complexities to promote health and cure disease.