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Genetics as a Medical Specialty

This is an especially exciting time in medical and human genetics. Medical genetics has achieved a recognized role as the specialty of medicine that deals with the diagnosis, treatment, and management of hereditary disorders. The idea that medical genetics is concerned only with the inheritance of trivial, superficial, and rare characteristics has given way to an understanding of the fundamental role of the gene in basic life processes. Medical and human geneticists are at the forefront of investigations into human variation and human heredity while also participating in and benefiting from rapid progress in molecular biology, biochemistry, and cell biology. In particular, the last decade of the 20th century and the beginning of the 21st century have seen the initiation of the Human Genome Project, an international effort to determine the complete content of the human genome, defined simply as the sum total of the genetic information of our species, encoded within each nucleated cell of the body. In partnership with all the other disciplines of modern biology, the Human Genome Project is already revolutionizing human and medical genetics by providing fundamental insights into many diseases and promoting the development of far better diagnostic tools, preventive measures, and therapeutic methods in the near future. When completed, the Human Genome Project will make available the complete sequence of all human DNA; knowledge of the complete sequence will, in turn, allow the identification of all human genes and, ultimately, make it possible to determine how variation in these genes contributes to health and disease.

Relevance of Genetics to All Medical Practice

Although medical genetics has become a recognized specialty, it has also become abundantly clear that

genetics provides important unifying concepts that permeate and unify all medical practice. To give patients and their families the full benefit of expanding genetic knowledge, all physicians and their colleagues in the health professions need to understand the underlying principles of human genetics. The existence of alternative forms of a gene (alleles) in the population; the occurrence of similar phenotypes developed from mutation and variation at different loci; the importance of gene-gene and gene-environmental interactions in disease; the role of somatic mutation in cancer and aging; the feasibility of prenatal diagnosis, presymptomatic testing, and population screening; and the promise of powerful gene therapies are concepts that now permeate all medical practice and will become only more important in the future. Thus, genetic principles and approaches are not restricted to any one medical subspecialty.

One aspect of medical genetics practice relevant to all of medicine deserves special emphasis: it focuses not only on the patient but also on the entire family. A comprehensive family history is an important first step in the analysis of any disorder, whether or not the disorder is known to be genetic. As pointed out by Childs, "to fail to take a good family history is bad medicine. . . ." A family history is important because it can be critical in diagnosis, may show that a disorder is hereditary, can provide information about the natural history of a disease and variation in its expression, and can clarify the pattern of inheritance. The diagnosis of a hereditary condition allows the risk in other family members to be estimated, so that proper management, prevention, and counseling can be offered to the patient *and* the family.

Disciplines within Human and Medical Genetics

Genetics is a diverse subject concerned with variation and heredity in all living organisms. Within this broad field, human genetics is the science of variation and heredity in human beings, whereas medical