Editorial

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Impact on genetic medicine

Beatrice Paradiso

Department of Neuropathology and Pathology Unit, NorthEast- Italy.

***Corresponding author:** Beatrice Paradiso, Department of Neuropathology and Pathology Unit, NorthEast- Italy, E-mail: Beatrice_p@gmail.com

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Abstract

Genetics will become truly integrated into medical practice when the genetic contributions to common disorders, such as diabetes or cardiovascular disease, are understood and can be used to improve the quality of care. Medical genetics has been formally recognized as a medical specialty in the United States only within the past decade. Initially, medical genetics was concerned with relatively rare single gene or chromosomal disorders, but with the sequencing of the human genome, genetics has become the driving force in medical research and is now poised for integration into medical practice. This article offers a perspective on the role of genetics in medical practice and how this role may evolve over the next several years. The author classifies the genetic contribution to medical practice into three categories: monogenic or chromosomal disorders, more common disorders due to a monogenic or single gene mutation, and common multifactorial disorders in which the interaction of multiple genes and the environment contribute to the cause of the condition.

Introduction

The author discusses the positives and the potential pitfalls facing primary care physicians and specialists as they incorporate medical genetics into their practices and points out some of the anticipated changes medical genetics will necessitate, such as an increased medical genetics workforce, including geneticists and genetic counselors. Genetic medicine is the integration and application of genomic technologies allows biomedical researchers and clinicians to collect data from large study population and to understand disease and genetic bases of drug response. It includes genome structure, functional genomics, epigenomics, genome scale population genomics, systems analysis, pharmacogenomics and proteomics. The Division of Genetic Medicine provides an academic environment enabling researchers to explore new relationships between disease susceptibility and human genetics.

Uses of genetics in medicine

Prenatal screening tests are the most widely offered genetic tests across North America, whereby fragments of placental DNA fragments drawn from maternal blood are sequenced for genetic abnormalities. In recent years, cancer therapy has focused on using tumor-specific antigens elucidated by sequencing as the targets of biologic therapies. For example, ado-trastuzumab is a monoclonal chemotherapy combination drug that has reduced the 3-year disease-free remission rate of HER2-positive breast cancer by 11.3% from the prior standard of treatment. It is exciting to hypothesize how the expanding role of genomics in medicine will impact our understanding and classification of disease. Perhaps purely clinical diagnoses such as trigeminal neuralgia, major depressive disorder, or atopic dermatitis will reform in light of underlying genetic origins. Ultimately, this will better classify our understanding of illnesses and improve treatment strategies and research. The ethics of storing identifiable genetic information, the rights of patients to knowledge of such data, and the potential effects on stakeholders at all levels of health care are additional complex issues. However, given the current funding status and international attention garnered by precision medicine and genomics, it will certainly have its place in the future of medicine. Cancer treatment is also set to benefit from genomic information to predict how an individual will respond to drugs (known as pharmacogenomics) and inform prescription of the appropriate drug or dosage. Pharmacogenomic applications extend into many areas of clinical practice; for example, in the prescribing of drugs such as antidepressants, analgesics and anticoagulants.

Applications of genomic information have led to:

An increased knowledge of diseases like cancer and heart disease.

An increased knowledge of rare diseases like cystic fibrosis and Huntington’s disease.

Advancements in genetic testing technologies like DNA sequencing.

Advancements in new drug therapies that are targeted or tailored to an individual’s genetic information.

Conclusion

Genomic sequencing technologies have drastically improved over the years which has driven down the cost of genetic testing, making it more accessible to individuals. Because of this, many laboratories have started to offer genetic testing for ‘healthy’ individuals that want to know whether or not they have may have a risk of developing a genetic disease that the family history is not providing evidence of. As discussed above, this type of testing can be performed to assess whether someone has a high hereditary risk of developing cancer or heart disease.