Priorities of Clinical Genetics in Translational Biomedicine

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Abstract

In order to understand the promise of personalized medicine, Translational Bioinformatics (TBI) research will get to still address implementation issues across the clinical spectrum. Here we present the state-of-the-art of clinical implementation of TBI-based tools and resources. Open areas of clinically-relevant TBI research identified during this review include developing data standards and best practices, publicly available resources, integrative systems-level approaches, user-friendly tools for clinical support, cloud computing solutions, emerging technologies and means to deal with pressing legal, ethical and social issues.

Introduction

During this capacity, TBI research and development efforts seek to enable the efficient and timely translation of biomolecular discoveries into actionable knowledge relevant to clinical end-points, so advance the state of human health and wellness. Although variations of “translational medicine” and “translational research” are widely wont to identify scientific programs in both public and personal organizations, when asked what the term means, even many would-be practitioners might respond with some version of the “I realize it once I see it” explanation. Our initial view was that the utilization of the term “translational” to explain this area of biomedical research is misleading, because the word carries many connotations, and is actually inaccurate.

Analysis

However, this view is debatable. The Encarta World English Dictionary defines translation as “the rendering of something written or spoken in one language in words of a special language. The part of this definition that applies to the concept of translational biomedicine is that the idea of various languages. Medical genetics is that the branch of drugs that involves the diagnosis and management of hereditary disorders.

Medical genetics differs from human genetics therein human genetics may be a field of research project which will or might not apply to medicine, while medical genetics refers to the appliance of genetics to medical aid. Medical genetics encompasses many various areas, including clinical practice of physicians, genetic counselors, and nutritionists, clinical diagnostic laboratory activities, and research into the causes and inheritance of genetic disorders.

Guidance is that the process of providing information about genetic conditions, diagnostic testing, and risks in other relations, within the framework of nondirective counseling. Genetic counselors are non-physician members of the medical genetics team who concentrate on family risk assessment and counseling of patients regarding genetic disorders. The precise role of the genetic counselor varies somewhat counting on the disorder.

Conclusion

Genetics has its roots back within the 19th century with the work of the Bohemian monk Mendel and other pioneering scientists, human genetics emerged later. It began to develop, albeit slowly, during the primary half the 20th century. Medical genetics was a late developer, emerging largely after the close of war II (1945) when the eugenics movement had fallen into disrepute. The Nazi misuse of eugenics sounded its death knell. there is a need for behavioral and social sciences to inform biomedical research for effective implementation. The third area speaks to the need for increased outreach and education efforts to improve the public’s genomic literacy such that individuals and communities can make informed health-related and societal (e.g., in legal or consumer settings) decisions. Finally, there is a need to prioritize representation of diverse communities in genomics research and equity of access to genomic technologies. Examples from National Institutes of Health-based intramural and extramural research programs and initiatives are used to discuss these points. The first area encompasses genetics of behavioral, social, and neurocognitive factors.

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