The expanding role of genetics in contraception and women’s health

Perhaps the most dramatic and important change in the practice of medicine over the past decade has been the integration of genetics into diagnostic protocols and therapeutic approaches. The Human Genome Project rightfully assumes considerable credit for this advancement; by marrying the resources of the United States government and private industry, incredible discoveries were made on an almost regular basis [1]. From the publication of the sequence of the human genome to the delineation of numerous mutations responsible for disease to a better understanding of the genetic mechanisms responsible for a variety of normal and abnormal physiologic functions, our understanding of biology, disease and genetic mechanisms have undergone a considerable change in the past decade. For many in science and medicine, these advances herald a new approach to the science and practice of medicine.

Science is obviously a critical component of our ability to provide effective care to our patients. Indeed, this explosion of genetic information has occurred in parallel with the integration of evidence-based medicine into conventional medical education and practice. As such, the use of scientific information to change the practice of medicine must be accompanied by a thorough understanding of the implications and potential effects of this information on patients and society in general, not just on the practice of medicine. An unfortunate example of the implications of ignoring the details of scientific information has been the response to the findings of the Women’s Health Initiative study (WHI). Clinicians worldwide began to consider hormone therapy to be an inappropriate therapy for menopausal women — all menopausal women. Although the WHI primarily studied an asymptomatic and older cohort, its findings were “assumed” to pertain to all women in menopause, even those who are younger and symptomatic. Indeed, it has taken almost 2 years for many professional organizations and clinicians to recognize the continuing important role of hormone therapy for the medical management of the symptomatic menopausal woman.

It is clear that scientific advancements alone cannot ensure improvements in the care of men and women; successful interpretation and implementation of such advances requires studies and educational programs to ensure that the new information will be appropriately applied in a manner acceptable to professionals and laity alike. In addition, society must provide effective safeguards to ensure that scientific advancements are not inappropriately used or manipulated to the detriment of large or small segments of society. Indeed, considerable funding (3–5% of the total budget) of the Human Genome Project was set aside for the study and evaluation of the impact of the Human Genome Project on health care and society [2]. It was widely recognized that the findings of the Human Genome Project would potentially alter the approach to preventive and conventional medicine. Premorbid identification of individuals at risk for developing disease could become possible along with the promise of preventative interventions and more effective therapies; however, such information could also be used in a variety of untoward situations, including denying insurance coverage and stigmatization of individuals [3]. A current example of both potentials is the concern of women who are at increased risk for developing breast and ovarian cancer based on family history about the impact of genetic (BRCA1/2) screening. The concern of these women and their clinicians is not just whether they have a mutation in one of these two genes that would result in a profound increased risk for developing breast and ovarian cancer, but whether their insurance provider would find out whether they have undergone screening. Currently, electing to undergo such screening could indicate a potential increased risk for these malignancies and thus could jeopardize a woman’s ability to obtain or maintain medical insurance coverage. In an effort to protect the confidentiality of their patients, many programs offer BRCA1/2 screening in an anonymous fashion or by offering to make available an alias. Nonetheless, such machinations deter many women from considering appropriate screening that could provide critical information concerning their risk and applicable surgical and medical management options and relegate women and clinicians to subterfuge and deceit [4,5].

Expanding our abilities to identify individuals at risk for developing certain diseases is a necessity if we are to improve our patient’s health and well-being. Encouraging reduced caloric intake and exercise will improve risk for adverse cardiovascular events. However, the delineation of genes associated with adverse cardiovascular events could considerably improve the benefits of those proven dietary...
and environmental interventions and determine the optimal preventative and therapeutic interventions for each individual. However, if such information would result in stigmatizing certain individuals based on the genetic complement and deny them certain privileges such as insurance coverage, then such scientific advances could be harmful to society in general [6]. Indeed, our current experience with BRCA screening shows that such discrimination already occurs in our health care system.

The provision of health care is rapidly progressing with 21st century technology, and yet, our health care system is mired in the 19th and 20th century bureaucracy. Insurability and access to health care must progress in a fashion commensurate with our technological advances. If the wondrous advances of the past few years are limited to only the wealthy and privileged, then these advances will have been achieved for minimal gain. Our professional missions may have different locations and languages and may impact a variety of social, racial, ethnic and economic groups; nonetheless, their goals are all the same. Their goals are to improve the world by caring for one person at a time. Indeed, it is written in the Talmud that to save one life is as if you have saved the world. However, to deny care to individuals because of a variety of personal, societal and economic factors, and now potentially upon genetic factors, is counter to our missions and a reversal of the Talmudic entreaty.

1. A proposal to ensure the successful marriage of genetics and medicine

Our scientific advances must be associated with new approaches to the ethical, moral and societal implications of these advances. Failure to implement such changes will at best create a disparate and dysfunctional system that provides ineffective care to all except the most wealthy and privileged and at worst will prevent the successful integration of these advances into health care [7]. However, it is foolhardy to think that solutions will be easy. Our initial goals should be simple and achievable.

- Continue to support the bench science that provides important scientific information and breakthroughs regarding the genetic and molecular aspects of health maintenance and disease.
- Ensure that such advancements are integrated into our health care system in an approach that takes into account the personal, societal and economic effects of such changes. Societal and governmental programs are already targeting the nonscientific implications of new scientific discoveries; support for such programs and publicity of their outcomes must continue if they are to facilitate the integration of new scientific advances into conventional health care.
- A reassessment of insurability must be undertaken with regard to the advances in genetics and molecular biology. The basis for determining insurability has historically been based on lifestyle issues such as smoking, obesity and motorcycle riding. With our increasing knowledge of the genetic component of disease, the insurance industry will have to incorporate this new information into the insurance paradigm but do so without discriminating against those with particular genetic sequences. Indeed, it is clear that the presence of certain genes and sequences will increase the risk of associated diseases and conditions but will not necessarily guarantee their phenotypic expression. In addition, delineating the presence of such genes allows for early, and possibly more effective, intervention and prevention.

We are in the midst of a great evolution in our understanding of the causes and mitigating factors in the development of disease. A few years ago, genetics had been the interest of a few molecular biologists and clinicians. Now, many primary-care clinicians and specialists consider their clinical and research work to involve genetic theory and practice. If we are to successfully incorporate these advancements into medical practice, we must ensure that this information will not be used to stigmatize and alienate large segments of our society. With the seminal role of insurance in the health care of our society, it is now time to strongly and forthrightly address this issue to ensure the facile integration of genetic advancements into our diagnostic and therapeutic practice, to prevent discrimination against whole components of our society and to improve the overall health care of all of our members of our society [7]. In a nation and society committed to the concept that all are created equal, failure to effectively study this major issue and implement rational and effective policies regarding insurability of all people will threaten the very core foundations that continue to ensure scientific advancements through our personal freedoms.

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References