

Short Communication

The Future of DNA Diagnostics

Mark H. Skolnick

*Myriad Genetics, Inc., Salt Lake City, Utah,
USA*

The cloning of BRCA1 [1] marked the beginning of the era of DNA diagnostics for common disease genes. Previously, genes had been cloned for rare genetic diseases, but not for genes which predispose to common diseases. The cloning of BRCA1 created a global awareness of the potential for DNA diagnostics to alter standard medical practice in appropriate situations.

A number of societal reactions surrounded this harbinger of sweeping change in health care practice. On one side, many individuals expressed concern over the potential for misuse of diagnostic information, especially by the insurance industry in the United States. Although such misuse has been noticeably absent, this concern had a strong positive influence on the creation of new legislation prohibiting such discrimination for most Americans. While some physicians chose to wait for this new paradigm to become a universal standard of practice, the American Society of Surgical Oncology has issued guidelines for appropriate testing for predisposition to breast and ovarian cancer, and many physicians embraced the opportunity to help their patients improve their quality of life and survival. Acceptance of genetic testing for predisposition to breast and ovarian cancer is becoming more widespread as the clinical utility of the test is reinforced by a series of studies, some of which are described in the accompanying abstracts. Although the existence of an important DNA diagnostic test is of

major importance per se, perhaps its greatest impact is as a signal that a new era in DNA diagnostics has begun.

There are two trends which must be considered to fully appreciate the probable impact of DNA diagnostics on medical practice in the next decade. First, there are now a number of genes being cloned which predispose to major diseases, including genes for prostate cancer, heart disease, diabetes, asthma, osteoporosis, obesity, and mental disorders. Many of these genes, when discovered, will have immediate clinical utility and will rapidly become important diagnostic tests. Others will be introduced more slowly, as further clinical or pharmaceutical research will be needed for them to have significant medical utility. Secondly, DNA diagnostic technology is in its infancy and should evolve dramatically over the next decade. Capillary sequencers, DNA chips, and mass spectroscopy are among the techniques under analysis to further automate DNA diagnostics which, in turn, will allow the technology to become widely utilized. The impact of technological change and more genes which have diagnostic utility has the potential to greatly alter medical horizons. Much greater emphasis will likely be placed on determining genetic predisposition, early diagnosis of disease and disease precursors, and the development of preventive therapeutics. These technical and medical changes should lead to great societal changes. This evolution of medical practice may be difficult for physicians, hospitals and agencies which must collectively adapt to new roles and a remodeling of the allocation of health care resources and delivery. However, I believe that the thrust of this new technology, which can dramatically alter the quality and length of many lives, will

drive this change. The political, economic and societal issues will necessarily be resolved so that DNA diagnostics can be fully integrated into a new era of medical practice.

References

- [1] Miki, Y. et al. A strong Candidate for Breast and Ovarian Cancer Susceptibility Gene BRCA1. *Science* **266**, (1994) 66–71.