Emergence of Genomic Medicine

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Genomic Medicine is a discipline in genetics that concern the recombinantDeoxyribonucleic Acid, Deoxyribonucleic Acid sequencing methods, and bioinformatics to series, assemble, and evaluate the purpose and organization of genomes. The accelerating development of biochemical and DNA-based diagnostic tests for human genetic conditions in the last decade has engendered a revolution in genetic diagnosis. Finally, the pace of development and application of DNA and biochemical genetic tests and their acceptance by the public may be accelerated by the recent widespread media coverage of the work of human geneticists.[1] A flood of large scale genomic and post genomic data means that many of the challenges in biomedical research are now the challenges in computational science.[2] Both genetic testing and genetic screening involve the same testing processes to examine an individual's chromosomes, DNA, or the biochemical product of a gene, typically a protein to confirm or refute a suspected chromosomal, DNA, or gene product change. Genomic medicine aims to revolutionize health care by applying our growing understanding of the molecular basis of disease. Progressively more, disease prediction, prevention and treatment will be tailored to each patient's unique genome, including the makeup of one's tumor or infectious microbes. All the fatal diseases that are threat to human life will be genetically modified in such a fashion to minimize the risk and cease the disease initiation like cardiovascular, cancer, endocrine diseases and infectious diseases etc.

Research in this arena is data intensive, which means data sets are large and highly heterogeneous. The identification of genetic disorders, and the potential for developing a therapy, is a powerful force in genetics and medicine. Diagnostic techniques can be used to identify specific proteins or fragments of DNA. The three basic components in genetic screening, that is, ethical, legal, and social issues, are to be considered and these genetic tests have to be performed with privacy, informed consent, and confidentiality. This brief discussion illustrates public expectations and fears about the effect of genomics, challenges to the goals of antidiscrimination laws and to the nature of the physician–patient relationship, and the contrasting perspectives and legal rules that apply to personal medical care and public health. [3]

Genetic testing for inherited genetic variants is performed for several purposes: diagnosis of individuals with symptoms, determination of future diseases. New testing technologies that will promote genetic testing in health care include DNA chip technology and tandem mass spectrometry. The current blood specimen collected from the heel of the newborn infant or blood collected from the umbilical cord would be tested by whole exome sequencing (WES) or whole genome sequencing (WGS), presumably within current New Born Screening (NBS) programs. Genetic screening would also expand NBS into nonmetabolic genetic disorders such as chromosomal abnormalities, neurofibromatosis, Duchenne muscular dystrophy, tuberous sclerosis, and many others. If screening included identifications of variations considered to increased risk for common diseases such as cancer, Alzheimer's disease, or Parkinson's, the infant would also be a proxy for family members leading to their testing for at-risk variations. Newborn genetic screening sounds like a "no-brainer." [4] Gene discovery is the goal of most contemporary human genomic research. Since the completion of the Human Genome Project, many new methods and tools for identifying disease susceptibility genes have become available, including haplotype-tagging single nucleotide polymorphisms based on the HapMap project28 and high-throughput technologies that allow examination of hundreds of thousands of genetic variants. [5] In light of this fact, a all-inclusive research agenda is desired to move human genome discoveries into health practice in a approach that capitalize on health benefits and minimizes harm to individuals and populations.

References

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