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Conceptual Paper

Genomic Medicine and its Future Prospects

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The enormous advances in genetics and genomics of the past decade have the potential to revolutionize health care, including mental health care, and bring about a system predominantly characterized by the practice of genomic and personalized medicine. The field of genetics has been recognized since the 19th century when Gregor Mendel studied his pea plants and discovered the fundamental laws of inheritance. But the term genomics was only coined in 1986, by the American geneticist Tom Roderick1 over a celebratory beer at a meeting about the mapping of the human genome. Genomics is now the accepted term for the study of the genome - the complete set of genes that encode for proteins, as well as the noncoding DNA in between - that exists in almost every healthy cell in the body (An exception is red blood cells, which do not have a nucleus so have no DNA, and no genome) [1].

The Human Genome Project (HGP) was an extraordinary, groundbreaking international research project completed in April 2003. It took 13 years and cost £2 billion. The HGP sequenced and mapped all of the genes that make up a human being, providing a complete genetic blueprint. The sequence is actually a composite derived from several individuals so it is a 'representative' or generic sequence. One use of the HGP is to look for genetic variations that increase risk of specific diseases, such as Alzheimer's disease, or to look for the type of genetic mutations frequently seen in cancerous cells. Technological developments now mean that the human genome can be sequenced in a few days for under £1000. Home-based 'spit tests' now available commercially online can provide limited genetic information for under [2,3].

How it works

The genome is one complete set of the genetic material in a cell. In humans, the genome is made up of around three billion nucleotide base pairs, which make up DNA molecules. Less than 2 per cent of the genome actually codes for proteins. The other 98 per cent is non-coding; some of the non-coding sequences regulate the transcription of proteins and some are transcribed to RNA but do not get translated into protein. It is a staggering fact that humans only have about 20,000 protein-coding genes - the same as a starfish. The function of the remaining non-coding DNA is not yet fully established, but it is thought that it may have a role in regulating and controlling gene expression. The genomic medicine is the use of information from the genome to guide clinical decision making. Personalized medicine is a broader concept that refers to a model of health care emphasizing the use of each individual's unique clinical, genetic, genomic, and environmental information for disease prevention and treatment. The state of science and technology is such that we can now examine and measure an individual's entire genome. Hence, individualized risk predictions and treatment decisions based on genomic information are theoretically possible, and in some instances, actually taking place. Personalized medicine draws on genomic medicine to leverage our knowledge of genetics and genomics for preventive health care, as well as administration of personalized, targeted therapies for individuals with existing conditions [4].

Use of genomics in medicine

Cancer

Testing both the patient's own genetic makeup ('germline' DNA) and the tumour DNA ('somatic' testing) are important in diagnosing and treating inherited cancers. 11 Only around 5 per cent of cancers are due to familial cancer syndromes such as breast or ovarian cancer, associated with mutations in the *BRCA1* and *BRCA2* genes. Testing allows members of these families to identify whether they are at increased risk and offers them options. Some may choose to do nothing; others will opt for increased screening or preventive surgery. Carriers of a gene mutation may opt for assisted reproduction (i.e. *In vitro* fertilization) with pre-implantation testing of embryos so that only unaffected embryos are used. This may sound a bit like eugenics to some, but in families that have been blighted by early cancer deaths for generations, these techniques offer a chance to eradicate further cases in future offspring. Some ethicists argue that once this chance to spare future offspring undue suffering exists, it is immoral not to use it [5].

Infectious diseases

Genomics is set to transform the way we diagnose and treat infectious diseases within the next five years. If we have a patient with tuberculosis, sequencing the genome of the microbe will be used to predict which antimicrobials are most likely to work. In public health, genomic information will allow tracking and planning of strategies to combat potential epidemics like swine flu and Zika. Public Health England has already used whole genome sequencing to track *Salmonella* out breaks [5].

Rare diseases

Rare diseases are not that rare overall they affect about 7 per cent of the population and are usually inherited, in over 80 per cent of cases. Detailed molecular diagnosis is important to allow proper management and follow-up as well as alerting other family members. Examples include cystic fibrosis, Huntington disease, some rare childhood cancers and polycystic kidney disease. The 100,000 Genomes Project is recruiting people with rare genetic disease who do not have a molecular diagnosis yet [5].

Common complex diseases

All aspects of health - good and bad - are a combination of genetic and environmental factors. Genetic profiling will identify susceptible individuals so that they can have screening and lifestyle advice tailored to their higher than average risk. This may be particularly useful in the case of prostate and breast cancer as well as

common conditions like diabetes. Many people do not know much about their family history - they may be adopted, have lost touch or family members may have died prematurely from other causes. Genomic profiling will give people a more accurate way of knowing whether they are at risk of common conditions. This carries its own problems: anxiety, the need to tell family members who may prefer not to know, the duty to inform insurance companies and distress if further screening or intervention is recommended but not available on the National Health Services [5].

Therapeutic Promise of the Genomics

With the completion of the Human Genome Project, anticipation was high that genetic information would radically improve medicine, that side effects would be more predictable, and that patients could be screened for likely drug responses. But thus far, progress has been much slower than what the initial excitement suggested.

A great deal of this delay relates to the fact that an individual's response to drugs is multifactorial, resulting from multiple gene and environmental interactions. Scientists also recognize that even as the knowledge base continues to expand, the clinical translation of that knowledge still requires empirical evidence, generated for a particular disease and drug combination, before treatment can be customized to a patient's genotype. Thus, much work remains to be done before personalized medicine can reach its fullest potential.

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