

A Note on Future of Genetic Medicine

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Introduction

Gene therapy will eventually play a major role in modern medicine. However, some authorities assert that if medical researchers move more cautiously and deliberately, society will benefit. Gene therapy is currently an elegant idea that has been clumsily implemented. That's not a criticism; it's just the way it is for a highly complex technology that is still in its early stages. After all, it has only been 5 years since the idea of gene therapy was conclusively proven to offer the X-linked severe combined immunodeficiency (X-SCID) condition at least a long-term therapeutic impact, if not a cure. In the future, gene therapy will be crucial to modern medicine. However, other authorities contend that society will profit if medical researchers go more methodically and carefully. Currently, gene therapy is a beautiful concept that has been awkwardly put into practise. That's not a complaint; it's just the way things are with a young, extremely complex technology. After all, it has only been 5 years since it was unambiguously demonstrated that gene therapy can, if not provide a cure for, at least provide a long-term therapeutic impact for the X-linked severe combined immunodeficiency (X-SCID) illness.

Description

Both patients are still alive and doing well now, however conventional therapy (pegylated bovine ADA, or PEG-ADA) administered prior to, during, and following their gene therapy confused the results and complicated any claim of "cure" based on the gene therapy. One patient's ADA level is 25% over normal, and 15% of her peripheral blood mononuclear cells carry the therapeutic gene. The therapeutic gene is barely detectable in the peripheral blood cells of the other, whose ADA level is less than 5% of normal. Anderson later participated in 12 of the first 20 gene therapy experiments that were authorised in the US. He commented, "In fact, within 20 years, I expect that gene therapy will be used regularly to alleviate — and even cure — numerous disorders," in an article regarding the potential of gene therapy.

Anderson was once again featured in Time's special genetics issue published to mark the 50th anniversary of Watson and Crick's discovery of the structure of DNA in 1953. Always upbeat, Anderson predicted to Time that "by 2053, there will be a gene-based cure for almost every disease." The prevalence of cancer, heart disease, and other contemporary ills will significantly decline. Numerous ideas about the purpose and reach of genetic testing are changing as a result of advances in genetic technology, which are having a significant impact in the clinic. Genomic testing increases the possibility of diagnosis or future diagnostic predictions, but it also increases the likelihood of ambiguous or unexpected results, many of which may have effects on several members of a person's family. In the past, genetic testing was rarely able to offer quick findings, but this is changing as genomic testing becomes faster and more widely available. As a result, genomic data is more

and more influencing decisions about patient treatment in the acute inpatient setting. Treatment options for genetic disorders are changing, which is having an impact on how doctors talk about formerly incurable diseases. Additionally, the point of access to testing is evolving due to an increase in direct consumer offering outside of the conventional hospital context.

Our technical capacity to sequence genetic information on a large scale has undergone significant changes during the last 20 years. In the past, genetic testing often consisted of either extensive genome-wide dosage screening at low resolution, such as karyotyping, or very detailed molecular testing of designated single genes. For example, the Human Genome Project, which was 99% complete in 2004, cost \$3 billion and took 13 years to sequence. Genome sequencing was too slow and expensive to be employed in clinical settings. Exome and genome sequencing have more recently made it possible to perform extensive genetic testing on a patient-by-patient basis within a clinically meaningful timescale. Exome tests sequence the entirety of the genome's protein-coding area, which makes up less than 2% of the total genome but is responsible for 85% of all known disease-causing variations; genome sequencing not only includes the exome but also all of the non-protein-coding DNA. Exome sequencing has been used as a clinical diagnostic tool more recently after initially being used in clinical research projects like the Deciphering Developmental Disorders project. Additionally, starting in June 2019, normal NHS tests will be able to order genome sequencing, which was previously only possible through programmes like the 100,000 Genomes Project [1-5].

Conclusion

The depth and breadth of sequencing technology have improved, and this has been crucial for better understanding cancer. The ability to sequence cancer genomes has facilitated the quick discovery of driver mutations and assisted in unravelling the intricate relationships between various cancer subclones over time and space, highlighting the enormous heterogeneity of cancers and the challenge of effectively treating them. It has been possible to identify previously unidentified mutational mechanisms, such as chromothripsis and kataegis, as sequencing tools have evolved to the point where small amounts of tumour or individual cells may be sequenced.

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