Evolution of a Field: The Role of Genetics in Modern Science

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Genetics is, most concisely, the science of inheritance and variation. It is the study of how characteristics are passed down from parent to progeny. This description, however, is misleading in its simplicity. During the twentieth century, genetics became a science which explains not only the nature of heritable material, but how this heritable material is expressed in organisms to determine how they develop and behave. There has been an extraordinary historical transformation in thinking within the discipline, instigated by the birth of molecular biology. The discovery of the structure of DNA illuminated the molecular structure of the Mendelian unit of heredity: the gene. This understanding of the chemical repository of genetic information, and the elucidation of the central dogma of molecular biology (DNA gives rise to RNA gives rise to protein), introduced a new, previously unknown language to scientists, and an explanation of the most fundamental chemical and physical mechanisms controlling life. It is the science of the mechanics of life. Genetics holds a unique central position in the biological sciences, despite the fact that genetic research is often undertaken by those who do not fall under the traditional classification of 'biologist'. Physicists, engineers, computer scientists and chemists can all be considered genetic researchers due to the nature of their work dissecting and utilising biological systems. The combined work of these scientists therefore places genetics at the forefront of modern science.

The genetic method for dissecting biological problems is unique. In combination with biochemical methods, we are able to dissect the pathways involved in the workings of the cell. In the twentieth century, "Genetics moves from a function, defined by mutation, to identify the gene responsible; physical interactions may be predicted from genetic interactions, and tested using standard molecular and biochemical techniques" [2]. The systematic analysis of mutants of a defined gene and the partners with which it interacts, and the phenotype that results, allows the elucidation of genetic pathways and the global interactions of genes within the genome. It is now possible to study genes and gene expression on a large scale, using high-throughput technologies. In the new century, 'DNA microarrays have revolutionised the analysis of gene expression by facilitating the monitoring of the RNA products of thousands of genes at once' [3]. Related fields include epigenomics (analysis of DNA modifications), transcriptomics (analysis of the complete collection of RNA transcribed in the cell), and proteomics (analysis of the complete set of proteins expressed).

As with genomics, these fields are most visible with regards their application to the identification of genes giving a predisposition for cancer, heart disease, obesity and conditions including depression and schizophrenia. All living organisms are a product of their genes and environment, and so it may become possible to identify the genes which influence personality traits, perhaps even sets of genes which give rise to traits such as impulsiveness or extroversion. It is now 8 years since the 'completion'* of the human genome project – although we have this 'blueprint of the human', we are still some way to understanding the complex interactions between all of the genes it contains.

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The discipline of genetics today is extremely broad, encompassing studies of the mechanisms of the workings of the cell (both prokaryotic and eukaryotic), to population genetics and studies of evolution and ecology. We benefit practically from our understanding of genetics in areas of healthcare, agriculture and, increasingly, engineering and materials science. Genetic research has led to the identification of the causes of hereditary and infectious disease – the field of clinical genetics is now a key area of healthcare, leading the way in the prediction and prevention of disease. The study of genetics has also given us the ability to genetically engineer organisms: we can program bacteria to produce an exogenous protein, develop pest-resistant crops, and modify and utilize cells for research into complex pathways. The technologies of cloning and genetic engineering can be applied to plant crops and agricultural livestock, allowing the identification and development of the most robust and desirable organisms for human use, improving the efficiency of food production – an important challenge with the world population having hit 7 billion last year [1]. Looking forward, cloning technologies could also lead to the recovery of endangered or extinct animals, breathing fresh life into the world's threatened biodiversity.

The genetic modification of crops is big business. In 2007, 9% of global primary crops were from genetically modified strains [4]. The most common plant cloning vector is the Ti (tumour-inducing) plasmid found in the bacterium Agrobacterium tumefaciens. This is a large, circular, double-stranded DNA molecule, which is able to replicate independently of the A. tumefaciens genome. A 30,000 base-pair segment of the Ti plasmid is able to separate from the plasmid and incorporate itself into the host plant cell genome. Insertion of gene sequences into this 'T-DNA' can be used to cause plants to express desired phenotypes [5]. The use of this, and related biotechnologies applied to crops, allow the production of crops which are not only resistant to herbicides, but genetically engineered to have a greater nutritional content and become hardier to environmental conditions including lack of water. It might also be possible to develop plants able to metabolise atmospheric toxins.

The fruit-fly *Drosphila melanogaster* is a commonly used, and historically important, model organism in genetics. Reproduced from [7]

The greatest amount of funding for genetic research is for the study of understanding and treating disease. The influence of genetic variation in disease may be the result of the possession (or lack) of one or more alleles which directly result in an abnormal phenotype, the combination of the possession (or lack) of particular alleles, and external factors such as DNA damage by external agents (carcinogens) or infection by pathogenic microorganisms. Medical geneticists specialize in the diagnosis and management of hereditary disorders; however research in genetics extends beyond those genetic disorders which are present before birth. Genetic counselling is a field encompassing the diagnosis of individuals suffering from genetic disorders, provision of medical and emotional support to these people, and providing advice to those individuals at a higher risk of generating chromosomal abnormalities or birth defects in their children. The understanding of cell death and proliferation is perhaps one of the most important elements of research in genetics. Cancer is a collection of diseases which are the result of unregulated cell division and is the leading cause of death in the developed world. Studies of the cell cycle via genetic methods allow the analysis of cell proliferation pathways and the identification of the molecular protagonists of unregulated cell division. Genetics is a key contributor in our search for the 'cure' for cancer.

Geneticists exploit their knowledge of prokaryotes to develop treatments against pathogenic infections, identifying the mechanisms by which pathogens disrupt normal physiology. Genetic techniques, allowing complex manipulation of prokaryotes via gene targeting and transformation (amongst others), allow us to harness bacteria as micro-factories; the most widely known example being bacterial production of insulin. More recently, a bacterium that digests oil has been patented but not yet used or approved for use. Such 'genetic engineering' is perhaps the most controversial modern scientific breakthrough. The new field of synthetic biology utilises biological machinery to make novel biological elements and systems not found in nature. The J. Craig Venter Institute last year revealed that the first ever synthetic organism had been made. *Mycoplasma laboratorium* is the result of an entirely artificially synthesised bacterial genome being introduced into another bacterial cell. The next landmark change in modern science will mean that we will no longer simply be investigating and researching the genetic machinery and mechanisms of organisms, but making use of, and exploiting our knowledge of, genetics to begin creating our own biological machines.

The borders between scientific disciplines are blurring in the 21st century, especially amongst the life sciences, and although we have not reached a point where genetics has ceased to exist as a separate discipline, as Sydney Brenner predicted in the early ninties [6], there is no doubt as to the central position the field inhabits. Scientists from all disciplines are today required to possess an understanding of the nature of DNA in the same way that they need an understanding of the nature of the atom. In this manner, genetics is at the very forefront of modern science. Genetics is the language of life, but we are far from fluent. ■

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References

* There exist multiple definitions of the "complete sequence of the human genome", however, according to the International Genome Project's definition; the human genome has been successfully sequenced [4].

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