

***In the 150 years since Darwin wrote *On the Origin of Species* our knowledge of biology and genetics has increased enormously, leading to vast improvements in health and life expectancy. How might further understanding in these areas affect healthcare 150 years from now?***

### **Sixth-form winning entry (Miranda Robbins, Perse Upper School)**

Now that 6 years have passed since the Human Genome Project constructed the sequence of the 20,000-25,000 genes in human DNA, scientists have an enormous task looking through each gene individually to find its specific role in the human body. This heralds a new age of genomics, which among many other projects is looking to how genes can cause, and in the future, how knowledge of genetics may pre-empt a disease.

#### **“Variation Under Domestication” (Chapter i of “On the Origin of Species”)**

Eugenics, or put more plainly, selective breeding in humans, was first presented by Sir Francis Galton a cousin of Darwin’s, in the 1880s. It is something that will never be considered without, at the very least, some controversy. Historically, eugenics has been used to breed out genes regarded as less desirable, whether they are the genes of convicts such as in Vermont in 1931, or the genes of an ethnic minority such as the Aboriginals in Australia at roughly the same time period. Although the practice of eugenics might seem reminiscent to a Nazi regime and is rooted in the past, eugenics is finding its uses in today’s society- reproductive technology and genetic diagnosis encompassing its modern visage.

Improvements in technology have already made it possible to sort sperm and screen embryos in order to determine the sex of the embryo as well as whether it is predisposed to develop certain genetic diseases. The use of IVF in order to choose the embryo inseminated, allows parents to artificially select their offspring, but also prevents selective pregnancy termination of foetuses that, seen from genetic testing, are highly likely to develop Down’s Syndrome, cystic fibrosis, and other diseases that can be detected in early stages. Currently the science for germ line therapy- modifying faulty sections of DNA- is illegal to perform on foetuses, but can be used in children and adults.

Germ line gene therapy has applications beyond merely replacing mutant DNA; in the near future it may be possible to screen embryos to tell parents the height, eye colour and possibly even the IQ of their child. With such temptations, will any parents be able to leave the characteristics of their child to chance? Indeed, the law is all that stands in the way of a future of “designer babies” and an official end to natural selection.

#### **(II) “Variation Under Nature”**

In discovering the role of each gene in the body, scientists will be able to discover how a mutation of a given gene can lead to a disease. From understanding the full process from mutation to disease, eventually scientists may be able to synthesize drugs that disrupt the process, stopping the disease from ever appearing. However, there are complications with this technique of treatment, for example, the genetic disorder gyrate atrophy affects the eye and eventually leads to blindness. The retina is damaged by an enzyme defect that causes a build up of the amino acid ornithine. This defect is caused by 35 different mutations in a single gene and so finding preventative drugs specific to each mutation would take an extremely long time to develop.

#### **(III) “Struggle for existence”**

Due to advances in genetic diagnosis, rogue genes might soon be mapped out so people can be screened and warned if they have genetic predispositions to certain major diseases, resulting in their lifestyles being altered accordingly. Although there are obvious benefits to being forewarned about the likelihoods of developing specific diseases, there are serious concerns that arise from this possible technology. If people are told they are at high risk of a disease, they are bound to face prejudices including escalated insurance costs, even

though a person could purely be a carrier of genetic traits and never exhibit symptoms themselves. Another problem this information could cause is selective breeding due to prejudice. If somebody found out their new partner had high risks of developing a disease such as schizophrenia, this could cause prejudice as to whether their partner is prepared to accept future responsibilities resulting from the surfacing of the disease, as well as consideration of whether the genes should be passed on to their offspring if both partners carry above average risk DNA.

#### **(iv) “Natural Selection”**

Personalising treatment is being made possible by the ongoing advances in technology such as high-throughput DNA sequencing analyzers and microarrays. In the future, oncologists are hoping to use direct molecular readouts from genetics (gene expression) and proteomics (proteome expression) in attempts to find the best technique to fight malignant tumors. Using the readouts, scientists can tell whether certain molecular pathways are present in a given person’s tumor cells. This could result in a personalized therapy targeting the active pathways found present in the cells. At the moment, we are in the early stage of research where tumors are being collected for experimentation for informational purposes, and so curing many people on an individual basis is a treatment that might only take place in the relatively distant future. Designing treatments to an individual would further decrease the fight a human needs to put up to survive a disease and unless there was complication in a patient, would stop one person being more likely to survive disease than another, resulting in natural selection becoming more ‘obsolete’.

#### **(v) “Laws of Variation”**

As well as inheriting genetic diseases from mutations in a gene, it is also possible to inherit gene mutations that lead to advantages against diseases compared with other alleles. Age-related macular degeneration can cause blindness in the long-term. Detecting variations in the genes related to the disease can reveal those at greatest risk from developing it. People who do not have protective variants have an increased risk by 100 fold.

#### **(viii) “Hybridism”**

Damage to mitochondria in cells by oxidants can result in mutations. These mutations can lead to disease and although no treatments for these disorders currently exist, the field of gene therapy looks as though it might have a gleaming future. Where current treatment tries to limit the mutation’s process that causes the disease, gene therapy ‘cuts to the chase’ and removes a degenerate gene, replacing it with a healthy one. A new branch of medicine called regenerative medicine is using the recent research not only to protect life process, but also reverse aspects of the aging process. Gene therapy was first attempted in 1970 but with little success. Even a decade later, efforts to utilize the treatment failed, but now gene therapy has been heavily researched and experimented and arranged into three clear mission statements:

- For monogenic diseases- genetic engineer can remove the harmfully mutated gene and replace it with a healthy one
- Mutate and kill an infected or cancerous cell
- Encourage the production of a protein by promoting secretion by other cells (e.g. Hepatitis C treatment by enthusing interferon secretion)

#### **(Xi) “Geographical Distribution: Physical Conditions”**

Human life expectancy has increased greatly over the past century and nothing is to say, with continued breakthroughs in medicine, this age cannot be increased further. Studies have already shown that the life span in animals can be extended using selective breeding and genetic engineering.

Isolated populations such as tribes found in remote mountainous regions have reported human life spans reaching up to 160 years on more than one occasion. Although the gene pool these tribes reproduce in are limited, it has been suggested that their shared environmental experiences are responsible for this abnormally large life span, rather than genes inherited to the lucky few. Scientists have estimated that a person's inherited characteristics are responsible for only 25-30 percent of their life expectancy; the remaining 70-75 percent are dependent on the environmental factors the individual is exposed to. In the future scientists might look to developing ways of monitoring an individual's 'personal environmental exposure' to decrease the damage done by this proportion in order to increase their overall life expectancy.

#### (xiv) "Recapitulation"

It is clear to see health-care in the past 150 years has improved substantially, due not only to breakthroughs in medical treatments but also technology. I have chosen chapter titles from 'On the Origin of Species' to use as section headings to illustrate how the fundamental points Darwin made are still relevant in science today and will continue to be applicable in the future.

I believe the pace at which discoveries have been made in genetics will continue to be sustainable in the future due to the vast amounts of knowledge recently made available by the Human Genome Project and the International HapMap Project. Treatments for certain genetically inherited diseases seem possible to develop from this information, with the possibility to not only fix one's DNA but also to improve it. Scientifically the limitations are likely to be conquered within the next few centuries, ethically, proposed possibilities are sure to be highly controversial and unrealistic to be accepted immediately by Society.

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