Villiers Park Educational Trust

AS Activity 1: Exploring the Human Genome
Suggested response

The Basic Facts

DNA and protein
The four letters in the DNA alphabet - A, C, G and T - are used to carry the instructions for making an organism. The order (or sequence) of these letters holds the code, just like the order of letters that makes words mean something. Each set of three letters corresponds to a single amino acid. There are 20 different building blocks - amino acids - used in a bewildering array of combinations to produce our proteins. The different combinations make proteins as different as keratin in hair and haemoglobin in blood.

The genome
The human genome is made up of 3 billion bases of DNA, split into 24 chromosomes.
This information…

- would fill a stack of paperback books 61 m high
- would fill two hundred 500-page telephone directories
- would take a century to recite, if we recited at one letter per second for 24 hours a day
- if spread out 1 mm apart, would extend 3000 km or about 7000 times the height of the Empire State Building

Human cells
The human body is made up of 100 trillion cells. Each cell has at least one nucleus, which houses the chromosomes.
There is 1.8 m of DNA in each of our cells packed into a structure only 0.0001 cm across (it would easily fit on the head of a pin)
If all the DNA in the 100 trillion cells of the human body was put end to end it would reach to the sun and back over 600 times
Most human cells contain 46 chromosomes: pairs of chromosomes 1-22, and a pair of sex chromosomes (females have two Xs; males an X and a Y). Sperm and eggs contain one of each chromosome

Genes and variation
Housed along each chromosome is a selection of genes. The human genome contains about 20 000-25 000 genes.
Mice also have about 20 000 genes; in the nematode (C. elegans), the number is around 19,000; in yeast (S. cerevisiae) there are approximately 6,000 genes; and the microbe responsible for tuberculosis has around 4,000.
Between humans, our DNA differs by only 0.2 per cent, or 1 in 500 base (letters). This takes into account that human cells have two copies of the genome.
Human DNA is 98 per cent identical to chimpanzees.

1. What does the sequencing of the human genome mean to you, your family, society, and eventually your descendents?
The DNA sequences generated in hundreds of genome projects now provide scientists with the “parts lists” containing instructions for how an organism builds, operates, maintains and reproduces itself while responding to various environmental conditions. But we still have very little knowledge of how cells use this information to ‘come alive’. The function of many genes remains unknown. Nor do we understand how genes and the proteins they encode interact with each other and with the environment. If we are to realise the potential of the genome projects, with far-ranging applications to such diverse fields as medicine, energy, and the environment, we must obtain this new level of knowledge.

One of the greatest impacts of having whole-genome sequences and powerful new genomic technologies may be an entirely new approach to conducting biological research. In the past, researchers studied one or a few genes or proteins at a time. Because life doesn't operate in such isolation, this inherently provided incomplete and often inaccurate views. Researchers now can approach questions systematically and on a much grander scale. They can study all the genes expressed in a particular environment or all the gene products in a specific tissue, organ or tumor. Other analyses will focus on how tens of thousands of genes and proteins work together in interconnected networks to orchestrate the chemistry of life, a new field called "systems biology".

Some possible impacts –

Molecular Medicine
Improve diagnosis of disease
Detect genetic predispositions to disease
Create drugs based on molecular information
Use gene therapy and control systems as drugs
Design 'custom drugs' based on individual genetic profiles

Microbial Genomics
Rapidly detect and treat pathogens (disease-causing microbes) in clinical practice
Develop new energy sources (bio-fuels)
Monitor environments to detect pollutants
Protect citizenry from biological and chemical warfare
Clean up toxic waste safely and efficiently

Risk Assessment
Evaluate the health risks faced by individuals who may be exposed to radiation (including low levels in industrial areas) and to cancer-causing chemicals and toxins

Bioarchaeology, Anthropology, Evolution, and Human Migration
Study evolution through germline mutations in lineages
Study migration of different population groups based on maternal genetic inheritance
Study mutations on the Y chromosome to trace lineage and migration of males
Compare breakpoints in the evolution of mutations with ages of populations and historical events

DNA Identification
Identify potential suspects whose DNA may match evidence left at crime scenes
Exonerate persons wrongly accused of crimes
Identify crime, catastrophe, and other victims
Establish paternity and other family relationships
Identify endangered and protected species as an aid to wildlife officials
Detect bacteria and other organisms that may pollute air, water, soil, and food
Match organ donors with recipients in transplant programs
Determine pedigree for seed or livestock breeds
Authenticate consumables such as caviar and wine

Agriculture, Livestock Breeding, and Bio-processing
Grow disease-, insect-, and drought-resistant crops
Breed healthier, more productive, disease-resistant farm animals
Grow more nutritious produce
Develop bio-pesticides
Incorporate edible vaccines into food products
Develop new environmental cleanup uses for plants like tobacco

2. If DNA passports are introduced who should be able to access this personal data? Is this type of passport a good idea? Why or why not?

Ideas to discuss -
Since its inception the Human Genome Project has dedicated funds toward identifying and addressing the ethical, legal, and social issues surrounding the availability of the new data and capabilities. Examples of such issues follow.

Privacy and confidentiality of genetic information. Who owns and controls genetic information? Is genetic privacy different from medical privacy?

Fairness in the use of genetic information by insurers, employers, courts, schools, adoption agencies, and the military, among others. Who should have access to personal genetic information, and how will it be used?

Psychological impact, stigmatisation, and discrimination due to an individual's genetic makeup. How does personal genetic information affect self-identity and society's perceptions?

Reproductive issues including adequate and informed consent and the use of genetic information in reproductive decision making. Do healthcare personnel properly counsel parents about risks and limitations? What are the larger societal issues raised by new reproductive technologies?

Clinical issues including the education of doctors and other health-service providers, people identified with genetic conditions, and the general public; and the implementation of standards and quality-control measures. How should health professionals be prepared for the new genetics? How can the public be educated to make informed choices? How will genetic tests be evaluated and regulated for accuracy, reliability, and usefulness? (Currently, there is little regulation.) How does society balance current scientific limitations and social risk with long-term benefits?

Fairness in access to advanced genomic technologies. Who will benefit? Will there be major worldwide inequities?

Uncertainties associated with gene tests for susceptibilities and complex conditions (e.g., heart disease, diabetes, and Alzheimer's disease). Should testing be performed when no treatment is available or when interpretation is unsure? Should children be tested for susceptibility to adult-onset diseases?

Conceptual and philosophical implications regarding human responsibility, free will vs genetic determinism, and concepts of health and disease. Do our genes influence our behaviour and can we control it? What is considered acceptable diversity? Where is the line drawn between medical treatment and enhancement?

Health and environmental issues concerning genetically modified (GM) foods and microbes. Are GM foods and other products safe for humans and the environment? How will these technologies affect developing nations' dependence on industrialised nations?

Commercialisation of products including property rights (patents, copyrights, and trade secrets) and accessibility of data and materials. Will patenting DNA sequences limit their accessibility and development into useful products?

3. What do you think about personalised disease management?

Human DNA is 98% similar to the chimpanzee, therefore how do we determine a personalised disease management programme or 'designer' treatment for each individual's genetic makeup. Slight variations in our DNA sequences can have a major impact on whether or not we develop a disease and on our responses to such environmental factors as infectious microbes, toxins, and drugs. One of the most common types of sequence variation is the single nucleotide polymorphism (SNP). SNPs are sites in the human genome where individuals differ in their DNA sequence, often by a single base. For example, one person might have the DNA base A where another might have C, and so on. Scientists believe the human genome has at least 10 million SNPs, and they are generating different types of maps of these sites, which can occur both in genes and non-coding regions.

Sets of SNPs on the same chromosome are inherited in blocks (haplotypes). In 2002 a consortium of researchers from six countries established a 3-year effort to construct a map of the patterns of SNPs that occur across populations in Africa, Asia, and the United States. Researchers hope that dramatically decreasing the number of individual SNPs to be scanned will provide a shortcut for tracking down the DNA regions associated with common complex diseases such as cancer, heart disease, diabetes, and some forms of mental illness. The new map also may be useful in understanding how genetic variation contributes to responses to environmental factors.