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nowgen
A Centre for Genetics in Healthcare

The Impact of Genetics on Medicine:
A Guide for Science Teachers

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A Guide for Science Teachers

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Nowgen, A Centre for Genetics in Healthcare, is a partnership project between Central Manchester and Manchester Children's University Hospitals NHS Trust, Lancaster University, The University of Liverpool and The University of Manchester. The Nowgen Centre is part-funded by the European Regional Development Fund and North West Regional Development Agency.

Equipment suppliers

BioRad - http://www.bio-rad.com
Select ‘Life Science Education’ and then ‘classroom kits ‘to view the equipment that Biorad have to offer. These kits are easy to use and designed to be fun in the classroom. Please be aware that some protocols may require a thermal cycler machine.

Edvotek - http://www.edvotek.co.uk
Edvotek manufacture a large number of kits covering a wide range of practical topics. The kits are designed to be used in the classroom but please be aware that some may require a thermal cycler machine.

NCBE - www.ncbe.reading.ac.uk
There are a range of kits available for schools from the National Centre for Biotechnology Education (NCBE), that come with excellent protocols. They have produced some simple electrophoresis tanks that are much smaller and cheaper than the ones used in research labs.

A medical revolution?

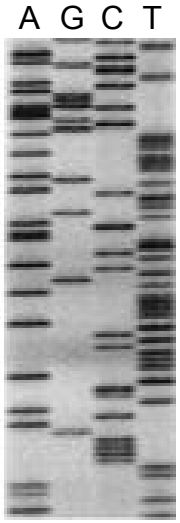
Human genetics research is advancing very rapidly. The post-genome era holds great promise and no area stands to benefit more than medicine. Many expect DNA testing to become a routine procedure within the health service. In future years genetic and genomic tests might be used to:

- **Diagnose more accurately**
  eg: identifying whether diabetic children can be treated by controlling their diet, without resorting to insulin injections
- **Predict responses to prescribed drugs**
  eg: using a genetic test to predict whether Herceptin will be effective in treating breast cancer
- **Investigate disease susceptibility**
  eg: identifying people who are more likely to develop an illness in the future, such as diabetes or heart disease

The potential is high, and expectations may be higher still, though questions remain about whether these aspirations are achievable, as well as consideration of ethics and equity. The impact of genetics is being felt already by patients, and the next generation is expected to feel its full force. Nowgen is working with teachers and A-level students to update them on the latest developments, and to explore their views on the future uses of genetic technologies.

The pace of discovery

- The structure of DNA was discovered in 1953.
- The first NHS genetic testing laboratories were set up in 1985, but very few diagnostic genetic tests were available until the 1990s.
- In 1975, Frederick Sanger invented a method to 'sequence' DNA. At this time, the method was used to sequence DNA of lengths up to roughly 300 base pairs and would have taken approximately one week.
- From 1990 onwards, teams of scientists took 13 years and $2.7 billion to sequence the first human genome.
- Now, ultra high-throughput genotyping machines can sequence nearly 4.5 million bases of DNA per hour. At this rate a human genome could be sequenced in a matter of days.



An autoradiograph showing Sanger sequencing

By using state-of-the-art molecular and computing technologies, and working collaboratively world-wide to share knowledge within the scientific community, the rate of progression in genetics is set to increase still. It is this rapid advancement in genetics, and associated technologies, that has the capacity to change healthcare as we now know it.

Genetic Interest Group - www.gig.org.uk/genesandyoucontents.htm

KS4, KS5 

The Genetic Interest Group (GIG) represents patients who have genetic conditions. This website provides summaries of these conditions, as well as lesson plans, ideas for stimulating activities and accompanying worksheets. Most importantly, GIG provides the patient's perspective on issues. The pack is called *Genes and You* and it can be freely downloaded from the GIG website. The site also provides links to other patient support group websites, many of which contain further information that could be used to provide a real-life context to teaching genetics.

John Kyrk - www.johnkyrk.com

KS4, KS5 

If you are searching for amazing animations, then this website below will bowl you over. Processes such as mitosis and meiosis, transcription and translation are played out, allowing viewers to visualise these mechanisms and to further understand how they relate to one another. This website is very carefully compiled, accurate and complex, therefore it is worth looking at to see how you could incorporate these animations into your lessons.

Genetic Futures - www.geneticfutures.com

KS4, KS5

Films can be a powerful way of stimulating discussion and we recommend one called *The Gift* which follows a family faced with making difficult decisions about genetic testing. The website gives you free access to short clips from this film (and others) and provides accompanying information about the issues. Alternatively, you can buy the DVD from the Y Touring Theatre Company's link on the website.

Genetic Science Learning Centre - http://gslc.genetics.utah.edu

KS4, KS5

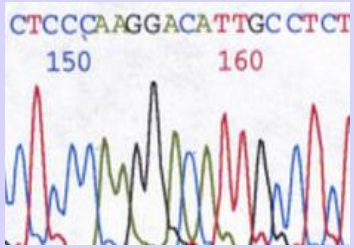
The Genetic Science Learning Centre in Utah, USA, has some fantastic resources. From their home page you can click on the *do try this at home* link to get instructions on how to extract DNA from plants and other tissues. The website provides clear instructions, and advice on extra investigations that could be used as extension activities. They have also produced some virtual experiments in *The Biotechniques Laboratory.* From here you can see animations detailing techniques such as DNA chips and electrophoresis. These are detailed and challenging, but give an accurate reflection of the processes.

Unravelling the genome

In 2003 the human genome sequence was announced. This breakthrough revealed the immense complexity of the human body. It is estimated that only 2-3% of the human genome sequence actually codes for proteins. Knowing the sequence of DNA allows scientists to begin to piece together information about how that sequence is transcribed and made into proteins, and how these processes are controlled and co-ordinated.

The genomes of many other species have been 'sequenced' alongside humans, and all the information is freely available on the internet. This knowledge allows scientists to compare humans to other species, termed 'model organisms', providing clues about how human genes function. Scientists are also using DNA sequence information to investigate the genetic variants that contribute to disease.

Case study: Sequencing individual genomes



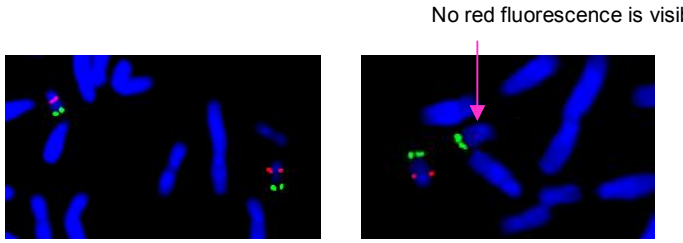
Modern DNA sequencing

James Watson, one of the team who solved the structure of DNA in 1953, had his genome sequenced and published on the internet in 2007. Interestingly, he refused to publish the sequence of his Apolipoprotein E gene, one mutation of which has been shown to increase the risk of developing Alzheimer disease. Sequencing James Watson's genome took 2 months and cost $2 million. Experts predict that technology capable of sequencing an individual human genome for $1000 may be available commercially in 2013.

Supporting people with genetic conditions

Traditionally genetics services in healthcare have focused on rare, single-gene conditions and how they affect individuals and families. Different methods of genetic testing are used depending on the condition. For example, for patients expected to have a large change in their DNA, scientists study chromosomes under a microscope. For smaller changes, such as the deletion of one or more genes, scientists might use a method termed FISH (Fluorescence *in situ* Hybridisation) where a piece of DNA will bind to a chromosome and fluoresce. If the gene is missing, there is no fluorescent signal.

The FISH technique



Unaffected individual **Patient with DiGeorge Syndrome**

FISH uses fluorescent DNA 'probes' to show whether small sections of chromosomes are deleted. The photographs above show a magnified view of two patients' chromosomes. The green probe is used to find chromosome 22 and the red probe is used to identify a specific section of chromosome 22. The left photograph shows a normal result, but the right photograph lacks the red signal, indicating that this section of the chromosome is deleted. This deletion causes DiGeorge Syndrome.

fun activities suggested within *Discovering DNA –The Recipe for Life* booklet, and *Case Studies in Cell and Molecular Biology* which provides detailed information on technologies such as cloning and gene therapy. These booklets can be freely downloaded, or sent to you on request.

BEEP - www.beep.ac.uk

KS4, KS5 

Bioethics Education Project (BEEP) is an excellent website designed to help teachers and students learn about bioethics. It provides a sound introduction to biomedical ethics and guidance for teachers on how to explore these issues with students. As well as covering a wide range of topics including pharmacogenetics, gene therapy, and cloning, this resource uses news headlines to provide a real-life context to class discussions. There are also links to interactive discussion forums for teachers and students, video clips and great ideas for lesson plans.

DNA Learning Centre - www.dnalc.org

KS4, KS5 

The DNA Learning Centre's website is extremely comprehensive and impressive. If you only have time to look in one place, this should be it. We recommend the Animations Library within the Resources section, and *Your Genes, Your Health* which is particularly helpful in explaining genetic conditions. The applications listed within *DNA Interactive* are very detailed and most suited to A-level students.

ABPI - www.abpischools.org.uk

KS3, KS4, KS5,

The Association of the British Pharmaceutical Industry (ABPI) website has a wealth of information covering many areas of science. We recommend *Biotech*, a resource pack for post-16 students which explores various technologies such as bioinformatics and DNA chips. *Biotech* can be freely downloaded from the website by clicking on to the teachers' section and then viewing the resources list.

@Bristol - www.at-bristol.org.uk/cz/teachers

KS3, KS4, KS5,

Citizen Science is a range of inventive resources that have been developed to engage young people in discussion about biomedical issues. We particularly like the materials in *Genome Games* that are free to download from the website above. They are ideal starter activities and get students talking by playing a game like Taboo or Pictionary. They are designed for students aged 11-16, but are fun for older groups too.

BBSRC - http://www.bbsrc.ac.uk/society/index.html

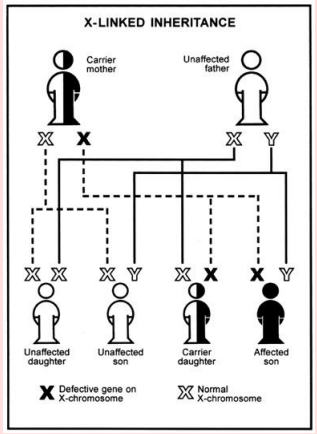
KS3, KS4, KS5

A number of excellent resources have been created for schools by the Biotechnology and Biological Sciences Research Council (BBSRC). We particularly recommend the

Genetic conditions can also be caused by changes to single bases in the DNA code. For example, a patient suspected to have cystic fibrosis can be screened for 31 common changes (mutations) in the CFTR (Cystic Fibrosis Trans-membrane Conductance Regulator) gene using a Polymerase Chain Reaction (PCR) method (a technique which produces millions of copies of the target DNA sequence).

There are presently over 1600 genetic tests available through the National Health Service (NHS) in the UK with this number set to increase significantly over the coming decades.

Case study: Empowering patients with knowledge



Inheritance pattern of DMD

Duchenne Muscular Dystrophy (DMD) causes muscles to weaken over time. It is caused by a mutation in the dystrophin gene, which is located on the X chromosome. Before genetic testing was available, women from families affected by DMD had no explicit diagnosis of whether they carried a mutated copy of the gene, which could be passed on to their children. In 1987 there were estimated to be 214 families affected by DMD in the North West of England, with 929 women at risk of being a carrier. In 2000, this figure had fallen dramatically following the introduction of genetic testing – with many women being told that they were definitely *not* carriers. Genetic tests are not always used to identify problems, they often reveal good news, offer reassurance and reduce people's risks.

A new era of genetic medicine

In recent years much of the research on human genetics has focused on complex, common diseases, rather than rare 'genetic' conditions. It is important to note that these complex diseases, such as diabetes and heart disease, develop mainly as a result of environmental factors, but genetic predisposition also plays a role. One of the most ambitious approaches in investigating these illnesses involves using biobanks.

Case study: UK Biobank

biobank uk

Improving the health of future generations

2007 saw the launch of the UK Biobank. This project aims to recruit 500,000 people between 40-69 years from all across the UK. The participants donate blood, provide information on their lifestyle, and allow researchers to access their health records in the future. Collecting banks of information on this scale will be incredibly valuable to researchers investigating the influence of genetics and lifestyle on people's health.
The ultimate goal of this major project is to improve the prevention, diagnosis and treatment of a wide range of illnesses (such as cancer, heart disease, diabetes, dementia, and joint problems).

Training for Teachers

By running workshops for teachers, Nowgen provides valuable continuing professional development in the rapidly changing field of genetics. We have organised a number of courses and often work closely with the Science Learning Centre North West.

For the latest information about teacher and student events visit: www.nowgen.org.uk

Recommended websites

KEY- Website includes:

Teachers' subject knowledge 

Ideas for experiments 

Facilitating debate 

Great animations 

Useful video clips 

Stimulating Students

Nowgen runs various workshops for A-level students and aims to excite young people about genetic medicine with engaging and thought-provoking events. In our state-of-the-art laboratory, we provide hands-on experience, giving an insight into genetics research. Nowgen have run workshops for over almost 4,000 young people to date.

Students enjoying the 'Hands-on' Molecular Genetics workshop at Nowgen, where they extract, PCR and analyse their own DNA samples.

Feedback from an A-level student who enjoyed being in the lab:
"It helps you to understand and relate to things you only read in textbooks."

Feedback from a teacher who brought a group of students:
"Excellent - innovative, interactive and kept them busy. Ofsted would have been impressed."

Personalised medicines

Genetic information can be used to predict how patients are likely to respond to specific drugs and drug doses. This field is termed 'pharmacogenetics' and is often referred to as 'personalised medicine'. Importantly, pharmacogenetics can prevent patients experiencing serious side effects, and will help the NHS provide more safe and effective treatments. Currently, there are only a few drugs that have genetic tests associated with them to see how patients will respond to them, but there are many more DNA tests that are expected to be used clinically in the near future.

Case study – Pharmacogenetics and Abacavir

Using genetics to identify the best medicine

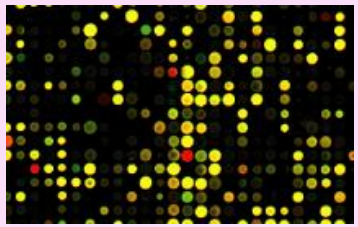
Abacavir is a drug that is used to treat HIV. In the past, 5-10% of patients would experience serious side effects when using this medicine, which in rare circumstances could be fatal.
In 2005, a genetic test was introduced into the NHS that identified people who were at risk of this side-effect. This test involved screening the HLA-B*5701 gene and people found to have specific mutations were prescribed an alternative therapy. The number of people experiencing side effects with this medicine has now dropped to virtually zero.

Understanding Cancer

Cancer cells develop because of DNA damage. The cells begin to replicate uncontrollably because the cell cycle is not being regulated properly. In a small number of cases, individuals are born with these changes in their DNA. Most cases of cancer are caused by damage to DNA during someone's lifetime (by environmental factors, eg: smoking). Cancer research often involves studying DNA and investigating the association between different genes and the disease.

Genetic tests are available for some patients (those rare patients who might have inherited a change in their DNA). These can be used to predict their risk of developing cancer in the future. If someone is found to be at high risk, doctors can reduce their risk by extra screening or potentially with surgery.

Case study: Using DNA chips to understand Cancer



A DNA chip

Technology such as DNA chips (also known as microarrays) can be used to aid the diagnosis of cancer. DNA chips allow scientists to understand which genes are switched on and are active in a tissue. For example, tumour cells from patients with Acute Lymphoblastoid Leukaemia, and Acute Myeloid Leukaemia are indistinguishable under a microscope. However, when microarrays are used to study the tissues in these tumours, they are clearly different from one another. Using this method for diagnosis informs the therapy that the patient is given. This type of testing is already used clinically in the USA and is expected to be used in the NHS within the next five years.

What is Nowgen?

Nowgen - A Centre for Genetics in Healthcare - is addressing the promise and challenge of genetic medicine. It is a not-for-profit organisation based on the Central Manchester and Manchester Children's Hospital Trust site, immediately next to The University of Manchester.

A multidisciplinary team undertake a range of programmes and research to improve clinical practice in genetic medicine. This team includes patients, artists, scientists, counsellors, health economists, ethicists, clinicians, educators and sociologists.

Manchester and the North West is a major centre for genetics research and we have the great advantage of having access to experts across the field of human genetics – from clinical geneticists to those pushing back the frontiers of scientific knowledge. Many of the leaders in these fields have already committed to getting involved with our education programmes.

A key area of Nowgen's work is to engage the public in dialogue, with a view to ensuring that mainstreaming genetic services in the NHS occurs in accordance with public opinion. We are keen to work with teachers and students to contribute to an informed and skilled citizenry who can respond independently and responsibly to emerging genetics in medicine.