Science and Society

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Genetic testing

English version

ELLS – European Learning Laboratory for the Life Sciences

EMBL

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1 Uses and the Risk of Abuse

Halldor Stefansson
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1.1 Genetic testing/Screening: areas of application

Areas of focus in genetic testing include: pre-implantation genetic diagnosis (PGD), prenatal diagnosis, newborn screening, carrier screening, susceptibility screening and forensic testing. These new identification technologies are presently being used for research and medical intervention, for reproductive information, for enumeration, monitoring, law enforcement and surveillance, and for registers of genetic diseases and disability.

Parents who are participating in *in vitro* fertilization programmes whose offspring are at potential genetic risk have special needs. **Pre-implantation** genetic diagnosis of embryos may ensure that only embryos free of genetic disease or problem traits will be placed in the uterus. **Prenatal diagnosis** discerns whether a foetus is at risk of various identifiable genetic diseases or traits. Prenatal diagnosis is commonly performed using chorionic villus sampling, amniocentesis testing, or ultrasound tomography. **Newborn screening** involves the analysis of blood or tissue samples taken in early infancy in order to detect genetic diseases where early intervention can avert serious health problems or death. This form of screening has been used widely to detect rare metabolic diseases such as phenylketonuria (PKU), sickle cell anaemia, and Tay-Sachs disease. **Carrier screening** identifies individuals with a gene or a chromosome abnormality that may cause problems either for offspring or the person screened. Groups tested include persons at risk, or a cross-section of the general public, for occurrence statistics. Such tests have been developed for cystic fibrosis, Duchenne muscular dystrophy, haemophilia, Huntington's disease, and neurofibromatosis, and the list is expanding. **Susceptibility screening** is not only used to identify inherited genetic mutations which lead to increased risk of developing a disease, for example, breast cancer susceptibility screening for BRACI and BRAC2 in women, it is also used to identify workers who may be susceptible to toxic substances that are found at their workplace and may cause future disabilities. And, finally, there is **forensic testing** where information obtained from genetic testing is used. It seeks to discover a genetic linkage between suspects and evidence discovered in criminal investigations. Test results are now commonly

presented as proof of innocence or guilt in court cases, and jury verdicts are commonly based on this type of genetic evidence. Critics note that forensic laboratories often test just once, unlike research laboratories which test many times, and that mistakes can be made. Confidentiality is also a major concern for many people in regard to genetic profiles obtained and stored in national police databanks. Innocent or guilty, more and more people are being subjected to new forms of invisible surveillance.

1.2 The social life of new genetic knowledge and technologies

The majority of people in our technologically advanced societies recognize the benefits and the potential usefulness of genetic testing and screening. However, the introduction of these new enabling technologies undeniably arouses various sorts of latent as well as manifest anxiety. In fact, as with all major new technologies, genetic testing and screening have their opponents as well as their proponents. Ruling authorities, both at the national and international level, have become increasingly aware of the social implications and polarization of perceptions in this area, and have introduced regulations and issued recommendations to allay collective discontent. These include: informing the public in advance; educating professionals to provide quality services (genetic tests should only be carried out by medically trained professionals); offering appropriate, non-directive, counselling; providing equality of access; respecting the self-determination of those tested; making testing or screening non-compulsory; and denying insurers the right to require testing or to seek the results of previous tests. The potential problems raised both by those who favour testing and screening, and by those who oppose it are similar. Those in favour think that regulatory or legislative solutions to the problems can be found while their opponents find the acceptance of the basic principles of eugenic theories (see below) and scientists' apparent inability to control outcomes of their genetic research as highly worrisome. They foresee a need to outlaw technologies that threaten privacy or civil rights and a need to protect against genetic discrimination. Disability advocates and feminists, for instance, have criticized genetic screening because they think it fosters intolerance for "less than perfect people".

1.3 Rethinking genetic determinism: Do our genes determine who we are?

Genetic determinism is the notion that our genes determine who we are at every level, physical, emotional and behavioural.

There are two schools of thought: 'Strong Genetic Determinism'—in other words, genes alone are sufficient to create appearances or behaviours, and 'Weak Genetic Determinism'—where genes play a role in appearance or behaviour, but other factors are involved as well.

In support of genetic determinism, the well-known biologist, Dean Hamer, has done significant research following the idea that our genes set the bar for our behaviour. His most controversial finding was the so-called "gay gene." It should be noted that nobody has been able to repeat the experiments that revealed the "gay gene". Moreover, genetic determinism has been used for the defence in murder trials. There are cases where violent criminals have been shown to have mutations in a gene associated with aggressive behaviour. However, the genetic 'evidence' presented to date has been over-ruled.

The ethical questions that arise are: Do genes make the person? How can you prove it? Is the environment alone responsible for how a person turns out? Which plays a bigger role? If evidence was found to support the "gay gene" is it really genes that primarily determine our sexual orientation? Do we need to take account of multiple factors that come into play determining who we are: family, school, television/media, life experiences, etc?

1.4 Genes and behaviour

Individuality is an evolving quality that emerges in, and from, human behaviour. Most people consider individuality to be heavily influenced by the human environment. The latter is always a complex and socially constructed artefact. However, recent molecular studies have exposed genetic factors that suggest a more biological origin for some forms of behaviour. Gene segments in the genome of humans and other animals have been identified and associated with particular behavioural traits. Is it possible that the presence or absence of even a single gene may predispose one to alcoholism, increased irritability and aggression, or enhanced intelligence? Clearly exploration of the nature versus nurture argument with regard to genetic predisposition has social, political and legal significance.

Employing "behaviour" as the experimental variant, however, requires identification of intrinsic behavioural characteristics that may be very difficult to define. Take 'intelligence'. It is considered an expression of behaviour, yet the delineation of what makes an individual intelligent has been highly debated. Does IQ determine intelligence? Or is economic success indicative of intelligence? Once an experimenter is comfortable with his proposed definition for 'a behaviour', 'intelligence', 'aggression', 'altruism', or whatever, the characteristic must be reliably and validly measured. However, if the relationship between, for example, aggression and a criminal record is not clear, then assigning parameters for levels of aggression will be even more challenging.

1.5 Our Eugenic past: The art of improving the Inborn qualities of a population (or of the human race)

In 1883, Sir Francis Galton coined the term "Eugenics". Eugenics refers to the doctrine which holds that the human race can be "improved" by selective control of breeding to eradicate less "desirable" traits in society. The supporters of eugenics

argue that social problems are caused by inherited genetic traits in people which can be bred out to resolve the problem for future generations. The logical conclusion of this theory is deeply racist and reactionary based on dubious research and prejudice. This led the to the introduction of sterilization laws in 27 States in the USA in 1931, and the sterilization of 350,000 people due to unwanted traits during the rise of the National Socialist Party in Germany in 1933.

Positive arguments for eugenics include the use of incentives and benefits to encourage the increased representation of certain genes in the gene pool of future generations. In other words, it is imperative that we give future generations the chance for the "best" possible start in life. More specifically, transplanting of sperm cells, and genetic testing of sperm and eggs could be seen as individual parents trying to fulfil their dreams for their children.
For supporters of evolution, eugenics is the next evolutionary step; the cost of living would surely decrease if there were fewer diseased people in the world (cheaper health care coverage, fewer taxes). The burden on society as a whole having to pay costs for institutionalization and/or hospitalization for those predisposed to various illnesses would be removed.

Negative arguments against eugenics include the possibility of Nazi-like sterilization, discrimination, killing of embryos, etc. The introduction of laws prohibiting marriages between those deemed "unfit" to produce offspring involves the elimination of unfit or undesirable genes by prohibition on sexual relations. We must ask ourselves if we are entitled to play the role of "The Creator"? Who is to decided on distinctions between desirable and undesirable traits to be introduced or eliminated from the common heritage of humanity? And in economic terms, will everyone be able to afford enhancement services in a neo-liberal, free-market society? Would the result be the advent of an "inferior" race/class, etc?

2 Genetic Counselling

Sabine Hentze
Human Geneticist

What genetic counselling can and cannot do

What if your father were diagnosed with Alzheimer's disease? What if there were a test that could tell you whether you, too, might develop the disease? How would the results of this test affect your marriage, your family, your job, your ability to receive medical insurance and other factors in your life?
A genetic counselling service can help weigh some of these questions, can help a person decide whether to undergo a test or not, and help them cope with the results.
The main function of a genetic counsellor is to provide clear, accurate information on the genetic, nature of a specific disorder. A counsellor should define the problem and frame it within the patient's medical history, family pedigree and other factors in his life.
A genetic counsellor will present the testing options available, the accuracy of the respective tests and discuss the experience with these tests. Tests can have consequences for an entire family—a spouse, siblings, parents, children, etc., and these need to be spelled out.

Undergoing a genetic test implies "**informed consent**" by the patient. The principle of informed consent means that tests which reveal personal genetic information are not mandatory. In other words, the patient has the right **not to know** and may choose ever to undergo a test, or never to access the test result.
One thing that a genetic counselling service cannot do is to instruct the patient on the course of action to take. However qualified and responsible genetic counsellors may be, they need to remain strictly neutral when consulted. Genetic counselling is to be, in other words, **non-directive**.
Whether or not to have DNA testing is an important decision for individuals and also for their families. Unfortunately, there is no right decision and no formula for making a decision of this type. There is no "one-size-fits-all" scenario. Each case is an intensely personal decision. This does not mean, however, that it has to be made in isolation, and that is where genetic counselling can play an important role.
Obviously, it does make a difference whether we discuss a genetic test that helps diagnose a metabolic disorder early, for which preventive care is available (e.g. hemochromatosis) or a severe disease like Huntington´s disease with progressive mental handicap where no cure is available.
Each disease presents its own special problems when it comes to counselling.

3 Cystic Fibrosis

Sabine Hentze
Human Geneticist

3.1 Cystic Fibrosis

Cystic fibrosis (CF) is caused by changes in a protein that controls the transfer of chloride and sodium ions (salts) across cell membranes. Disruption of salt transfer results in abnormal gland secretions and dehydration due to increased loss of salt and water during sweating. CF affects almost all of the glands in the body that secrete fluid, resulting in a variety of symptoms. Secretions may be thick and cause blockage in the pancreas, intestines and lungs. This mucus cannot be cleared from the lungs and provides good nutrition for bacteria, so affected children suffer from recurrent infection. In the pancreas the digestive enzymes do not reach the gut so the children show poor digestion, dehydration, do not grow properly and puberty may be delayed. Adults show more serious complications such as collapsed lung, heart failure, frequent infections and reduced life expectancy.

3.2 Genetics

CF is caused by mutations in the cystic fibrosis conductance transmembrane regulator (CFTR) gene on chromosome 7 which codes for the protein that controls ion transfer across cell membranes (Fig. 3.1). Molecular analysis has identified approximately 1000 mutations in the CFTR gene. Different mutations determine the severity of symptoms seen in CF patients.

3.3 Inheritance

Autosomal recessive, carrier frequency is 1: 20.

3.4 Incidence

CF is the most common hereditary disease leading to death among Caucasian people in the United States. The incidence of the disorder is: 1 in 2,500 Caucasians; 1 in 14,000 African Americans; 1 in 11,500 Hispanics; and 1 in 25,000 Asians.

3.5 Diagnosis without genetic screening

Half of the patients with CF are undiagnosed during the first year of life and 25 percent remain undiagnosed by the end of the second year.

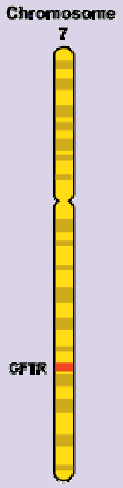

Fig. 3.1 CFTR on Chromosome 7

3.6 Clinical outcome without screening and treatment

If not diagnosed early, 13 percent of newborns and infants will die. Most untreated CF individuals will not live past their late 20's.

3.7 Clinical outcome with screening and treatment

If diagnosed and treated, newborn and infant mortality is reduced. Half of the people with CF live longer than 28 years due to availability of an increasingly wide range of treatments.

These include physical therapy, enzyme replacements, supplemental salt, antibiotics to control infection, oxygen therapy, surgery, rarely organ transplantation. Treatment for CF costs an average of 35,000 Euro per year per patient in direct medical costs alone. Gene therapy is under investigation and evaluation.

3.8 Testing

Mutation in the CFTR gene results in an increase in an enzyme called trypsinogen. The initial newborn screen tests for this enzyme use a dried blood sample. However, this is not a conclusive test for CF. Measuring the salt levels in sweat can usually confirm the diagnosis. Today, clinical suspicion of the disease is confirmed by molecular analysis.

3.9 Genetic counselling

Carrier screening of the general population is possible using DNA mutation analysis. CF carrier testing is not recommended at this time unless a family history of CF is present. However, it is performed more frequently in countries with a high disease incidence (e.g. Great Britain). The gene test will detect 85% of carriers. Newborn screening is discussed to prevent malnutrition as well as to stabilize lung function. Prenatal diagnosis is possible for most families. Embryo screening for CF is possible for carrier parents prior to embryo implantation in countries where Pre-implantation diagnosis is available.

4 Fact Sheet on Violent Behaviour

Giovanni Frazzetto
Branco Weiss Fellow

A link between genes and violent and aggressive behaviour has not been definitively identified, but various studies in mice and humans have shown a possible relationship between this type of behaviour and the expression of genes involved in molecular circuits in the brain.

One gene in question is located on chromosome X and it codes for an enzyme called monoamine oxidase A (MAOA). In the brain, MAOA metabolizes neurotransmitters such as norepinephrine, serotonin and dopamine, rendering them inactive.

Genetic deficiencies in MAOA activity have been linked with aggression in mice and humans.
Laboratory studies have shown that adult mice lacking the MAOA gene have increased levels of the neurotransmitters listed above and are more aggressive than the wildtype. If MAOA expression is restored in the same mice, aggression is normalized.
The first suggested evidence in humans came from a 1993 report of a Dutch family. Several men in this family had a defective MAOA gene. None of the enzyme was found in their brain and they were prone to impulsive bouts of aggression. This mutation is very rare and no other examples of this kind have been reported.

Scientists recently turned to larger studies checking the MAOA genotype of a broader population. A team of British scientists screened a group of New Zealand children who had been followed since birth (see Science article). A polymorphism for the MAOA gene exists. This means that individuals in the same population can have slightly different versions of the same gene and, therefore, different versions of the proteins they encode. Not all of these differences have crucial consequences.
In the MAOA case, scientists found that the polymorphism of some individuals gave rise to a version of the enzyme which had lower activity. This corresponds to higher levels of neurotransmitters in the brain and to higher incidences of aggressive behaviour. But having the right genotype turned out not to be enough!
Previous circumstantial evidence had suggested the hypothesis that childhood maltreatment predisposes one strongly to adult violence.

The New Zealand study established a neat interaction between the MAOA genetic background and environmental influence (maltreatment). In the study, environmental influences were critical. In the absence of abuse, individuals having the low-activity genotype did not show antisocial or aggressive behaviour.
This is a very good example of how social factors can play an enormous role in the expression of behavioural traits.

Establishing a genetic component in aggressive behaviour might well have legal implications. A piece of genetic information on an individual convicted for violent behaviour could easily influence a judge's decision.

5 Background Information: Genetics

Giovanni Frazzetto
Branco Weiss Fellow

Our genes are scattered throughout our DNA, which is organized in the cell nucleus into long ribbon-like structures called **chromosomes**[1]. Chromosomes come in pairs and humans have 23 pairs—46 chromosomes in total. 22 of the pairs are so-called autosomes. The 23rd pair is the sex-determining gonosomes: females carry 2 "X"-chromosomes, males one "X" and one "Y"-chromosome. The chromosomes of each pair carry the genes for the same traits, so each gene comes in 2 copies. They are found at the same place, or **locus**, on the chromosomes. However, they do not necessarily carry the identical information; e.g. we have 2 gene copies for the trait "eye colour". One copy could code for the colour "brown", the other for "blue". We call each copy of a gene an **allele**, and this is important in understanding genetic diseases.

However, nature "plays around" with the sequence of genes producing slight alterations in the sequence. They might have no functional effect (we call those variations "polymorphisms"). If they alter the gene and thus the protein it encodes and its function we call it a "mutation".

A gene with a mutation often causes disease. How serious this is depends on the function of the gene. It also depends on the **pattern of inheritance** of the feature linked to it.

Fig. 5.1 An artistic representation of human chromosomes, painted by Jacques Deshaies (2002)

[1] **Boldfaced** words have an entry in the mini-glossary.

The basic patterns of inheritance were discovered towards the end of the 19th century by the Austrian monk, Gregor Mendel, while he was studying the pattern of inheritance of some traits of peas plants. Although Gregor Mendel did not have the notion of 'gene', he postulated that characteristics were transmitted by hidden 'determinants' in the organism.

The early 20th century saw great progress in the fields of genetics and heritability, which formulated basic rules to explain how traits and features are passed from parent to child. The discovery of DNA as the basis of heredity and methods to analyse the sequences of genes have allowed scientists to link many disorders to mutations in single genes. This has given researchers good insights into the patterns by which genetic disorders are passed down. The enormous amount of information obtained on genes and their structure and organization has given physicians valuable tools for diagnosis.

Below you'll find a short description of different patterns of inheritance. While this list refers mainly to disease, the same patterns are responsible for the transmission of healthy features such as hair colour and blood type. These patterns work for autosomes. Sex chromosomal inheritance is more complex, as some genes on the X-chromosome may or may not follow the patterns described below.

Autosomal dominant pattern

This type of disorder occurs

- when the two homologous chromosomes carry different versions of a gene;
- one version is defective;
- it is sufficient to override the activity of the other, healthy copy;
- and this creates an abnormal **phenotype**, or physical characteristic of the body.

Brown eye-colour follows a dominant inheritance pattern. An example of autosomal dominant disorder is Huntington's disease. It is caused by a mutation in the so-called huntingtin gene on chromosome 4 which affects the central nervous system. Symptoms of this disease usually appear in adulthood (both in males and females).

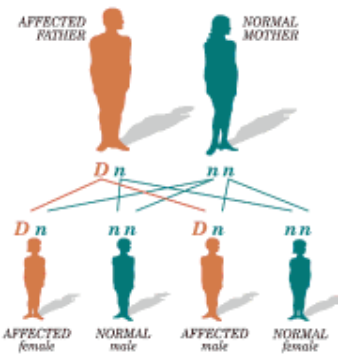

Fig. 5.2 Autosomal dominant pattern

Autosomal recessive pattern

This type of disorder is in some ways similar to the last pattern: homologous chromosomes carry different versions of a specific gene. However, in a recessive pattern, the healthy gene can override the dangerous effects of its mutant twin. The person does not develop the disease, but is a 'carrier' and there is a 50-50 chance that each of his children will inherit the defective copy. Carriers normally don't display any of the symptoms of a disease and are healthy.

The child of two carriers may inherit two copies of the recessive gene from both parents, and will suffer from disease.Recessive diseases include sickle-cell anaemia (affecting red blood cells) and cystic fibrosis (affecting the thickness of the mucus in lungs and pancreas). The inheritance of blue eyes follows the same pattern.

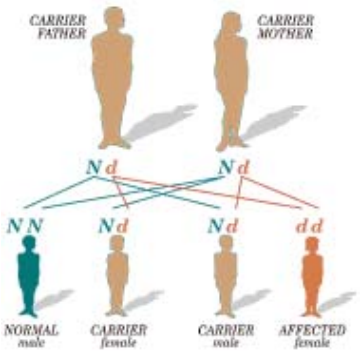

Fig. 5.3 Autosomal recessive pattern

X-linked inheritance pattern

This refers to disorders in which the defective gene lies on the X-chromosome. Females have two X-chromosomes, while males have an X- and a Y-chromosome. Mutations may be dominant or recessive (see above), but males only have one copy of the X gene. So X-linked disorders are most common in males: there is no second X-chromosome, and no functioning copy of a gene to compensate for the defect. For a recessive x-chromosomal gene females are healthy carriers. There is a 50% risk for the son to inherit the defective gene copy and develop the disease. Colour blindness, hemophilia A and Duchenne muscular dystrophy are examples of this.

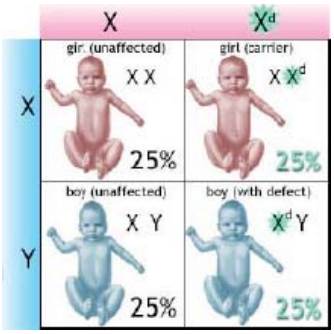

Fig. 5.4 X-linked inheritance pattern

Single gene and multi-factorial disorders

So far, these patterns cover disorders that arise from a flaw in a single gene. However, there are probably thousands of multi-factorial disorders.

Single gene disorders are brought about by a mutation in one specific gene, meaning that one or both copies of a gene pair are not functional. Examples of this are Duchenne muscular dystrophy, sickle-cell anaemia and neurofibromatosis.

Multi-factorial disorders come about when a disease arises from the combined effects of several gene pairs and environmental factors. The genes involved are called **susceptibility genes**, and the more genes involved in a disorder, the more complex patterns of heredity will be. Often diseases of this kind result from a complicated interaction between genetic and environmental factors, such as diet, smoking habits, exposure to radiation, chemicals, etc. Diabetes and some types of cancer fall into this category.

6 Mini-Glossary

Giovanni Frazzetto
Branco Weiss Fellow

ALLELE:
one of a set of alternative forms of a gene. In a diploid organism a gene has two alleles, each occupying the same position (locus) on homologous chromosomes.

CARRIER:
an individual who carries a copy of a recessive gene.

CHROMOSOME:
structure composed of a very long DNA molecule and associated proteins that carries the hereditary information.

GENETIC REDUCTIONISM/DETERMINSIM:
a philosophy establishing the primacy of nature over nurture by suggesting that human lives and actions are inevitable consequences of the biochemical properties of their cells and makes the genes possessed by an individual the essence of humanness.

KARYOTYPE:
full set of chromosomes of a cell arranged with respect to size, shape and number.

LOCUS:
the position of a gene on a chromosome. The term locus is usually restricted to positions of genes, which are expressed.

MULTI-FACTORIAL DISORDER:
A disorder which is brought on by the joint action of multiple factors. The contributing factors include several different genes as well as various types of agents from the environment.

MUTATION:
Any permanent change or alteration in the genetic material that changes the nature of the product made under the direction of that gene.

POLYMORPHISM:
It refers to the allelic variation of a gene within a population. Individuals in the same population can have slightly different versions of the same gene, and, therefore, different versions of the proteins they encode. Not always these differences have crucial consequences.

SINGLE-GENE GENETIC DISORDER:
A disorder which comes about when there is a mutation in a specific gene, and one (for a dominant disorder) or both (for a recessive disorder) of the genes in the gene

pair cannot function properly.

SOMATIC MUTATION:
A mutation which occurs in any of the body cells of an individual over the course of that person's life. Since the mutation is not in the eggs or the sperm cells, it cannot be passed on to children.

7 Background Reading_

Genetics and human behaviour the ethical content
Published by the Nuffield Council on Bioethics (2002):
http://www.nuffieldbioethics.org/genetics-and-behaviour/genetics-and-behaviour-chapter-downloads

- Chapter 9: Antisocial behaviour
- Chapter 14: Legal responsibility
- Chapter 15: Testing and selection in employment, education and insurance

Carson RA, Rothstein MA (1999) Behavioral Genetics: The Clash of Culture and Biology John Hopkins Univ Press

Convention for the Protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine:
Convention on Human Rights and Biomedicine, Oviedo, 4.IV.1997:
http://conventions.coe.int/treaty/en/treaties/html/164.htm

8 Useful Websites

The Genetic Interest Group in London:
www.gig.org.uk

DNA Gene Testing - Links and Resources:
http://insideout.rigb.org/ri/dna/notes.html

A website on Huntington's disease:
http://www.hda.org.uk/

A website on thrombophilia:
http://www.fvleiden.org/

Fragile X-Syndrome:
http://www.fragilex.org/

Websites on cancer:
http://www.cancer.org
http://oncolink.upenm.edu/disease/breast/
They contain general information on cancer, diagnosis, treatment, genetic testing, support groups and patient privacy.

Websites on cystic fibrosis:
http://www.cftrust.org.uk/index.jsp
www.cysticfibrosis.com

Activity – The Virtual Expert Committee

This activity is a role game that you can adapt for your classes. It involves court cases that revolve around issues of genetic testing.

The group will split into two expert committees (approx.15 people each). Each committee is asked by a court to provide a recommendation on a specific legal case involving genetic testing.

The committees will be composed of people from different walks of life, each of whom has had an experience with genetic testing and represents a specific interest group: a member of a family affected by a genetic disorder, a doctor, a politician, a lawyer, an officer from an insurance company and a state employer.

We will introduce you to two case scenarios illustrating potential societal and ethical conflicts raised by genetic testing.

Each member of the team will play one of the roles above, and you will be assisted by a moderator. “Role cards” will help you slip into the mindset of the person you are playing (see Appendix I and II). Remember, you need to stay in character with the role you have assumed in the game—expressing that person’s opinion rather than your own. At the end of a discussion with the other members of the committee you will need to reach a decision and provide the court with a recommendation as well as the reasons behind it. It is likely to be difficult to reach a consensus because each of you will have very different views.

However, try not to be shy and enjoy the game!!!

CASE STUDY I

Cystic fibrosis and prenatal diagnosis: Hughes vs. Medicis Insurance Company

Ben Hughes is three-years old and has cystic fibrosis (CF). Hospital admissions and treatment have so far cost more than $165,000. His private insurance company, Medicis, is only partly covering the expenses. The company refuses to fully cover the expenses arguing that the parents underwent a gene test before birth and knew the risk. Ben’s parents have sued the Insurance Company.

(Refer to fact-sheet on CF and Dr. Hentze).

Characters: (see Appendix I)

- Parent of a child affected by a genetic disorder
- Officer from a health insurance company
- Doctor (a clinical geneticist)
- Lawyer
- Man on the street
- Moderator

CASE STUDY II

Genetic testing for aggressive behaviour: Matt Scott vs. NY Police Academy

After joining the New York Police Academy, Matt Scott was informed that he would have to undergo a genetic test to check for violent behaviour susceptibility genes. Matt feared he could test positive. He felt that the test was discriminatory and refused to undertake it. The Academy claimed it had the right to choose their future officers carefully in order to avoid unjustified episodes of aggression and fired him. Matt claimed he had the right not to take the test and he sued the Police Academy.

(Refer to the Fact-sheet on violent behaviour and News of the Week).

Characters: (see Appendix II)

- Civil servant
- Employer
- Doctor (a clinical geneticist)
- Lawyer
- Officer from Amnesty International
- Man on the street
- Moderator

Appendix I: Role Cards for Case Study I

Doctor

You are a specialist in genetic disorders and you have been dealing with diseases affecting adults and children. Your point of view is that genetic testing will assist doctors in providing fast and accurate diagnosis and better medical care. You are also concerned about the importance of teaching the public about genetic disorders and to inform them of the advances that medicine has made in this field.

These ideas are crucial to your point of view:

• The importance of informing patients of the treatments available for each disease.

• You are concerned with the sensitivity of each test. You give high importance to the principles of genetic counselling.

Officer from a health insurance company

Insurance companies base the rates they offer to potential clients on a person's current or future health status and on the risks associated with their life style. The information of people's susceptibility to develop some kind of illness or disorder in the future may be crucial in your decision on the premiums they would have to pay. Remember you don't have a scientific background.

Some of your thoughts:

• Companies and employers should be allowed to demand genetic tests or to have access to results of tests that have already been done.

• Early identification of people with genetic disorders can lead to cost savings in treatment or care.

• Your company offers a service in exchange for money; if your business is no longer profitable, you will no longer be able to offer the service. Additionally, a very precise knowledge of risks helps you to offer better prices to all of your clients.

Family member

Your role is to voice the concerns of the parents of children with genetic disorders. You have an insider's view of the social perception of genetic diseases.

Some of your concerns:

• You would like to know which tests are available and how sensitive they are.

• Will genetic testing lead to more efficient treatment/cure of the disease?

• Who decides who will have access to the test results?

• Will the test information result in discrimination against individuals with genetic disorders?

Lawyer

You are concerned about protecting the rights of the individual and the rights of the employer.

You are aware that advances in genetic technology have raised many legal issues. Although you would appreciate general regulations, you grasp for a balanced solution for each specific case.

Some of your considerations:

• Should parents have the right to refuse screening?

• Each citizen's privacy is valuable and must be protected by laws.

• We need to weigh the economic interest of the individual and of the insurance company.

Man on the street

You are a citizen, who, like all of us, may one day be affected by issues raised by genetic testing.

You are a kind of joker in the game, since you do not represent any specific interest group and you can look at the case from many different perspectives.

Just be yourself!

Appendix II: Role Cards for Case Study II

Doctor

You are a specialist in mental and behavioural disorders. Your viewpoint is that genetic testing will assist doctors in providing fast and accurate diagnosis and better medical care.

However, you are also sceptical about the value of genetic tests in the prediction of behaviour.

Some of your concerns:

• The importance of informing patients of the treatments available for each disease.

• You are concerned with the sensitivity and reliability of each test.

• You also think genetic tests cannot give a holistic view of a person and cannot completely predict his/her behaviour.

Officer from Amnesty International

You are very concerned about incidences of police brutality and the use of excessive force. You have evidence of cases of police beating, unjustified shootings and the use of dangerous disciplinary tactics.

Some of your points:

• You think too little has been done to combat or avoid police brutality.

• In addition, from your perspective, racial and ethnic minorities have often disproportionately been the victims of police misconduct, including physical abuse.

• In general, you would like to prevent cases of aggression and would like to see regulations enacted that would be effective.

Civil Servant

As the policeman in our case you represent the voice of state employees, concerned that genetic information could be used against them in their working environment.

• You think you have the right not to take such tests and the right not to acquire genetic information about yourself

• You are sceptical about the real value of genetic tests

• You think they cannot tell everything about a person

Employer

As an employer you are in favour of genetic tests that could give you information about your employees' current and future health or conduct.

• You are in favour of tests that could predict whether your employees would be able to do their jobs

• You think genetic testing for employees should be a requirement.

Lawyer

You are concerned about protecting the rights of both individuals and employers.

You are aware that advances in genetic technology have many legal implications. Although you would appreciate general regulations, you believe that there needs to be a balanced solution for each specific case.

Some of your considerations:

• Should employees have the right to refuse screening?

• We need laws to protect citizens' privacy.

• We need to weigh the economic interest of the individual and that of the hiring company.

Man on the street

You are a citizen, who, like all of us, may one day be affected by issues of genetic testing.

You are kind of a joker in the game, since you do not represent any specific interest group and you can look at the case from many different perspectives.

Just be yourself!

Acknowledgements

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