Genetics and Health

~ SCIENCE 24 ~

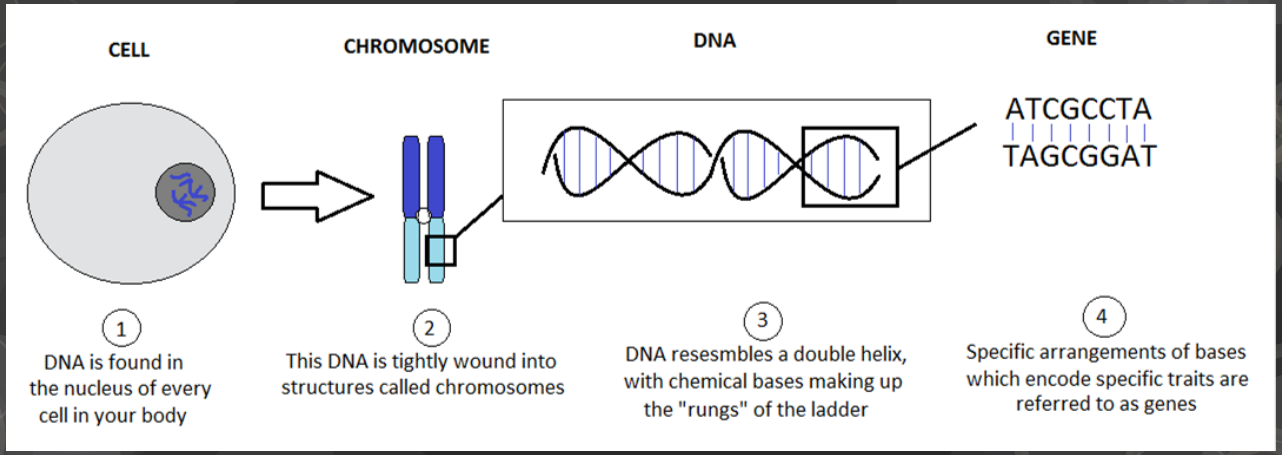
Inherited Characteristics

Have you ever been told that you look like your mom or dad? This is the result of inherited characteristics, or *genetics*

- Genetics is the study of how heritable traits are passed down from parents to offspring
- You receive 50% of your DNA from your mom, and 50% from your dad

Genes, DNA & Chromosomes

The *genes* you inherit from your parents are simply different arrangements of DNA which encode specific traits:



Chromosomes & Sex Determinatio

Every cell in the human body contains 23 pairs of chromosome (or 46 in total):

- One of these pairs are the sex chromosomes; that is, they determine whether an individual will be male or female
- Females have two identical “X” chromosomes, while males have one “X” and one “Y”
- Does the karyotype to the left represent a male or female?

DNA Replication

When your cells divide, the DNA in their nucleus must be replicated so the new cell contains the exact same genetic information.

- When DNA replicates, it opens up like the zipper of a coat
- Free floating bases within the nucleus then match up with the original strand to make an exact copy
- Adenine (A) always pairs with Thymine (T), and Cytosine (C) always pairs with Guanine (G)

(video)

DNA Replication & Mutation

The process of DNA replication is not a perfect one; mistakes are oft made, resulting in changes to genes and the way genes are express

- These changes are called *mutations*
- Mutations are not always “bad”; they often just result in variations of particular traits (e.g. blue eyes instead of brown)
- Mutation occurs naturally, but it can also be caused by exposure to dangerous substances, called *mutagens*
- Examples of mutagens include cigarettes, exposure to UV radiation, alcohol & other drugs

Acquired Genetic Disorders

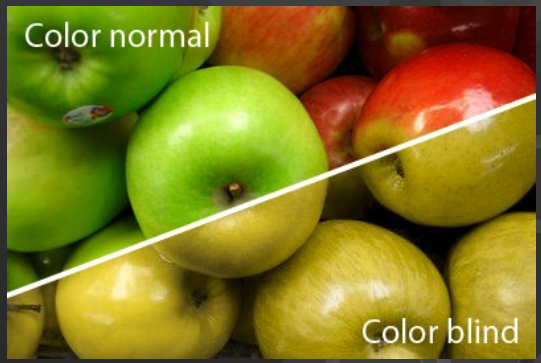
Some diseases can be acquired, or developed, throughout an individual's lifespan as a result of exposure to mutagens.

- Overexposure to the sun or UV rays from tanning beds, for example, can lead to skin cancer
- UV rays can alter gene expression, causing cells to divide uncontrollably

Heritable Genetic Disorders

If mutations occur in the *gametes* of an individual (e.g. during the formation of sperm or eggs), their offspring may inherit these disease-causing genes

- Colour blindness, for example, is caused by a mutation to the gene that encodes specific receptors in the eyes
- An individual who is colour blind may pass down this gene to their offspring



Patterns of Inheritance

Each of our genes has at least two possible variations; for example, t
for eye colour may encode blue eyes or brown eyes. Each variation, *allele*,
must therefore be either dominant or recessive.

- Because chromosomes come in pairs (one from each parent), we have two copies (alleles) of each gene
- However, because we receive one allele from each parent, both of our alleles for a given gene do not have to be exactly the same
- So how do you know which variation will be expressed?

Dominant Vs. Recessive Genes

Dominant variations of genes, or dominant *alleles*, will always be expressed over recessive variations of genes, or recessive *alleles*. Capital and lowercase letters are used to represent whether an allele is dominant or recessive:

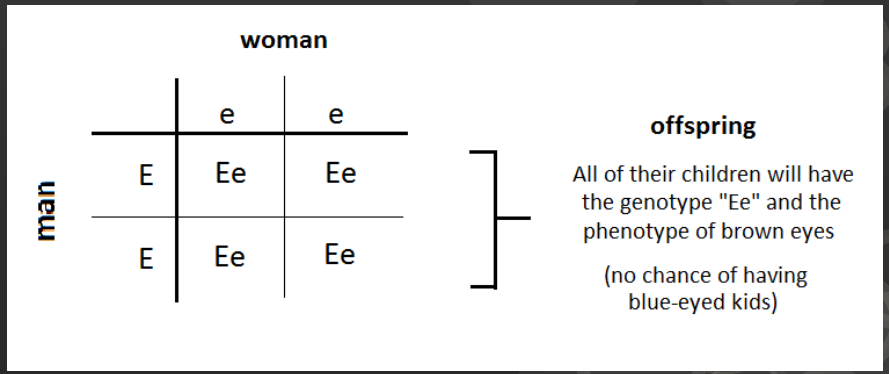
- E.g. the allele for brown eyes (E) is dominant over the allele for blue eyes (e)
- So, a person with one dominant allele (E) and one recessive allele (e) will always have brown eyes
- A person with two dominant alleles (EE) will always have brown eyes
- A person must have two recessive alleles (ee) to have blue eyes

Punnett Squares

If we know the *genotypes* (combination of dominant & recessive alleles) of tw
people for a particular trait, we can determine their probability of having a chi
each corresponding *phenotype* (observable characteristics) using a Punnett Squ

EXAMPLE:

A brown-eyed man with the genotype EE ("homozygous dominant") has children with a blue-eyed woman with the genotype ee ("homozygous recessive"). What is their chance of having a blue-eyed child?

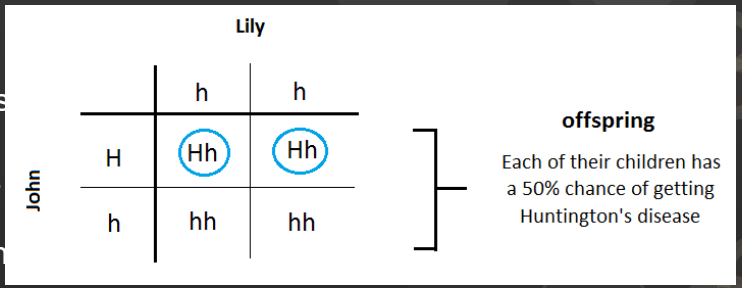


Genetic Counselling

A genetic counsellor uses Punnett Squares to predict whether or not the offsp
two individuals will inherit a particular genetic disorder:

EXAMPLE:

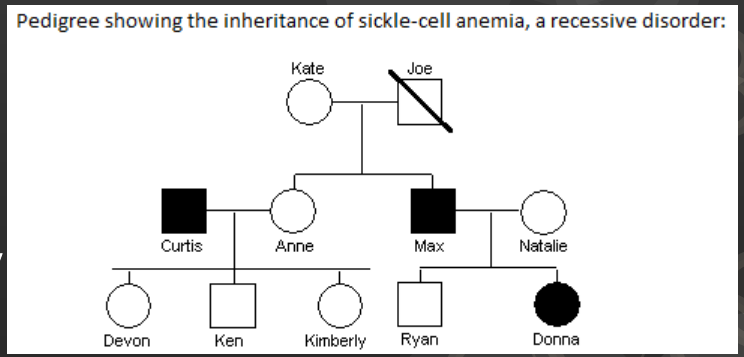
Huntington's disease is a dominant genetic disorder resulting in the degeneration of motor and cognitive abilities over time. John was diagnosed with Huntington's las year; with genetic testing, his genotype was determined to be Hh (heterozygous). John's wife, Lily, does not have Huntington's, so her genotype must be hh What is their probability of having a child with Huntington's?



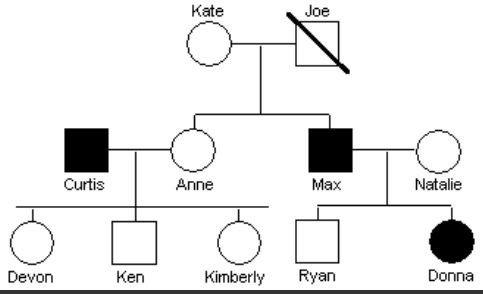
Pedigrees

A pedigree or “family tree” can be developed to represent the passing on of particular traits throughout each generation.

- females are represented using circles, males are represented using squares
- affected individuals are shaded in
- deceased individuals are crossed off
- successive generations are joined by vertical lines
- horizontal lines show mating



Pedigree showing the inheritance of sickle-cell anemia, a recessive disorder:

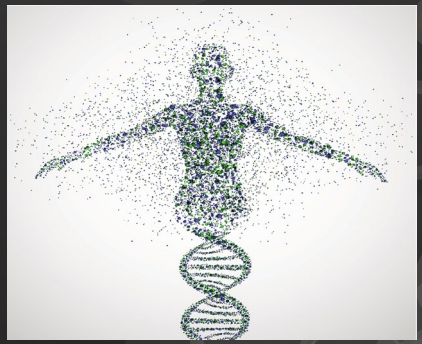


- Who are the children of Kate and Joe? *Anne and Max*
- Who is Max married to? *Natalie*
- Who is Ken's sister? *Kimberly*
- Who in the family has sickle-cell anemia? *Curtis, Max and Donna*
- What must be the genotype of Curtis, Max, and Donna? *aa*
- What could Anne's genotype be? *AA or Aa*

The Human Genome Project

In 1990, scientists began mapping the human genome. They hoped that by iden
all of the genes that make up the human genome, they would gain a better
understanding of what makes us so unique; they also hoped it would allow the
prevent, treat, and/or cure a number of diseases. By 2003, the project was com
They found that…

- The human genome consists of approximately 20,500 different genes (about the same number as a field mouse)
- Most of these genes do not appear to have any specific purpose (once called “junk” DNA)



Epigenetics: Decoding the Genom

Although the completion of the Human Genome Project was a huge accomplis in the field of genetics, it left many questions unanswered. Why are there so seemingly useless genes? How can we have the same amount of DNA as simpler organisms? It seemed as though the genetic code alone was not 1 responsible for making us who we are...

- In recent years, it has been discovered that "junk" DNA actually has a purpose; it is responsible for turning on & off other genes
- Environmental factors, including diet, exercise, the consumption of drugs & alcohol, and even mental health can attribute to the "flipping" of these epigenetic switches

E.g. women who experience anxiety during pregnancy are more likely to have children who are anxious as a result of epigenetics

Genetic Engineering

Our growing knowledge of genes and gene expression has resulted in leaps o innovation in the fields of medicine and agriculture through *genetic engineering*

- Genetic engineering is the process of taking a gene from one species an inserting it into the DNA of another. This may cause the organism to produce more or less of a particular gene, or express an entirely new tra altogether.
- Genetic engineering has allowed us to grow crops in regions they wouldn normally be able to grow; it also allows us to grow bigger, healthier, more nutritious crops
- Genetic engineering has allowed us to produce large amounts of insulin people with diabetes; it is also the basis of many other medical breakthroughs in terms of disease treatment and prevention

Ethical Concerns

Although an understanding of the human genome and the ability to alter it to improve our own health through genetic engineering has endless benefits, there a number of ethical concerns associated with such technologies...

- Who should be allowed to have access to our genetic information? Should a person (e.g. significant other, employer, insurance agency) be allowed to make decisions about another person based on their genetic information?
- Do companies have the right to patent genes?
- Should parents be able to genetically select the characteristics their offspring?
- If you had a fatal genetic disorder, would you want to know?