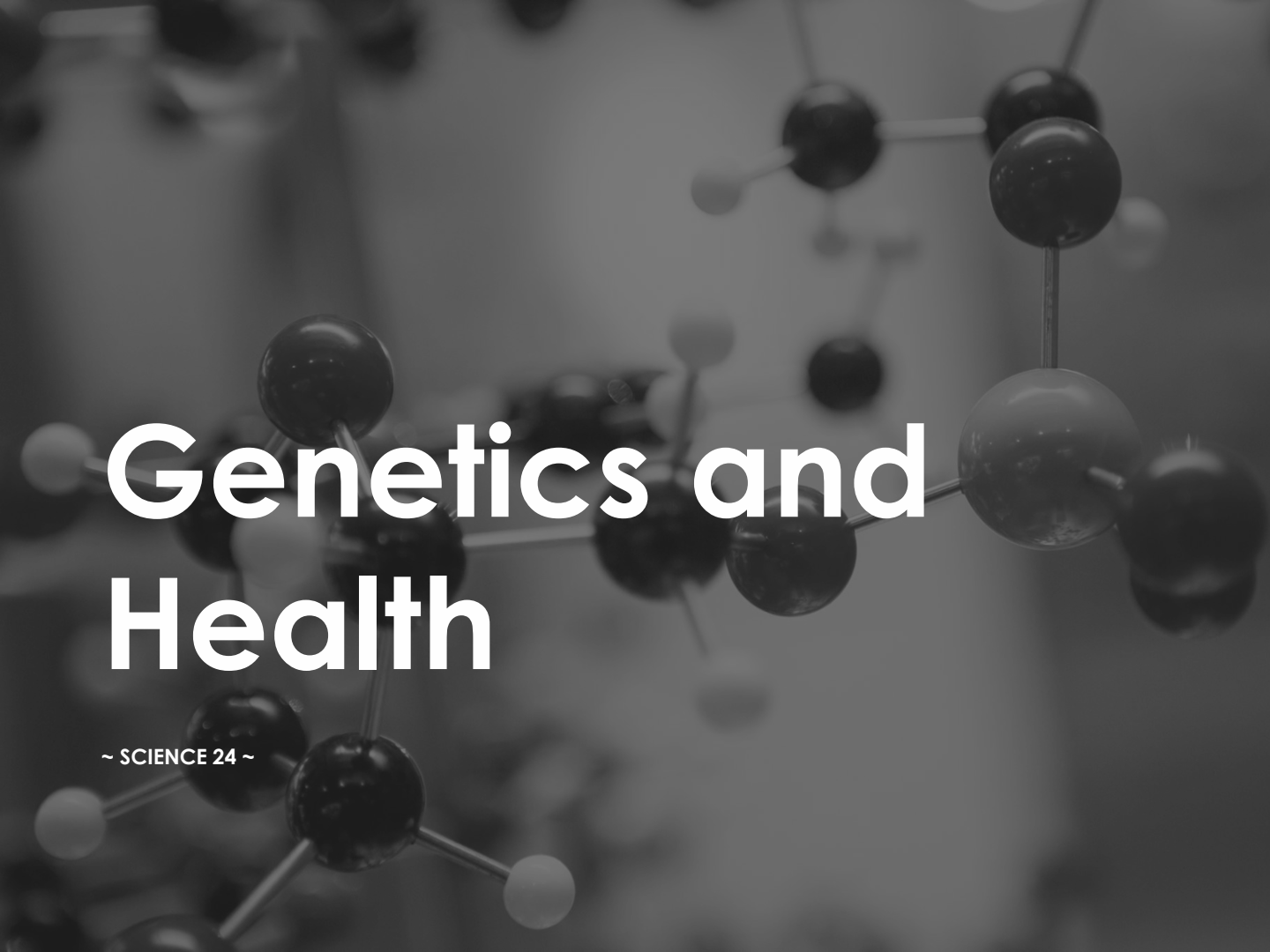


Genetics and Health

~ SCIENCE 24 ~

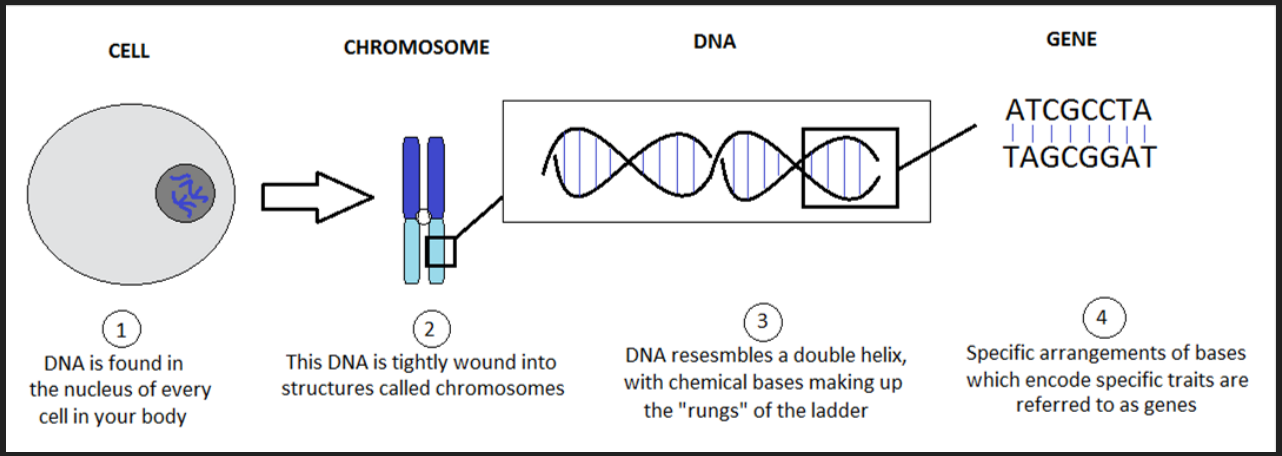
Inherited Characteristics

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

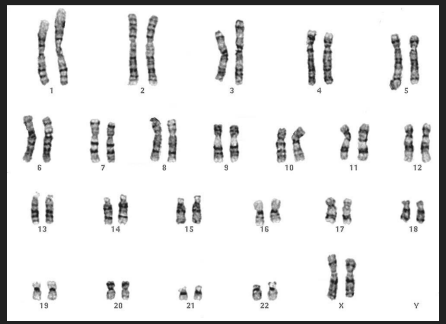
Genes, DNA & Chromosomes

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):

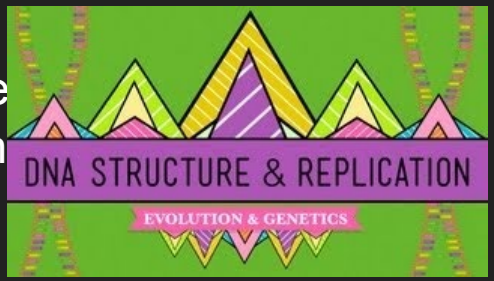


- One of these pairs is the sex chromosomes; they determine sex
- Females have **two** identical "X" chromosomes, while males have **one** "X" and one "Y"
- The chromosome map to the left is a **karyotype**, organized **largest to smallest**

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 compounds called "base 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)



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

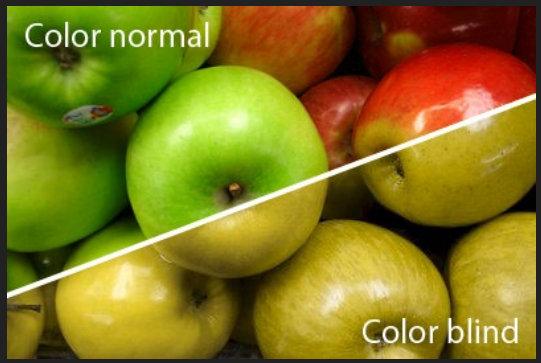
Some diseases can be acquired, or developed, throughout an individual's lifespan as a result of exposure to mutagens

- Over-exposure 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

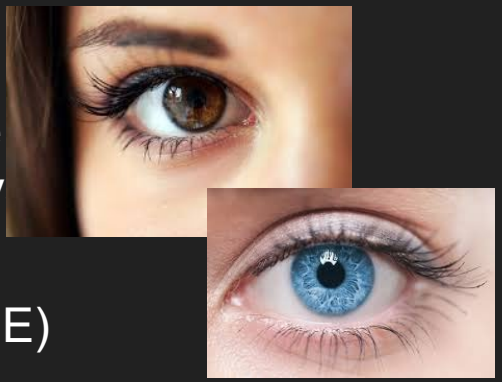
Each of our genes has at least two possible variations; for example, t
for eye colour may encode blue eyes or brown eyes. Each variation, *allele*,
must therefore be either dominant or recessive

- 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 hav 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
- So, a person with one dominant allele 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

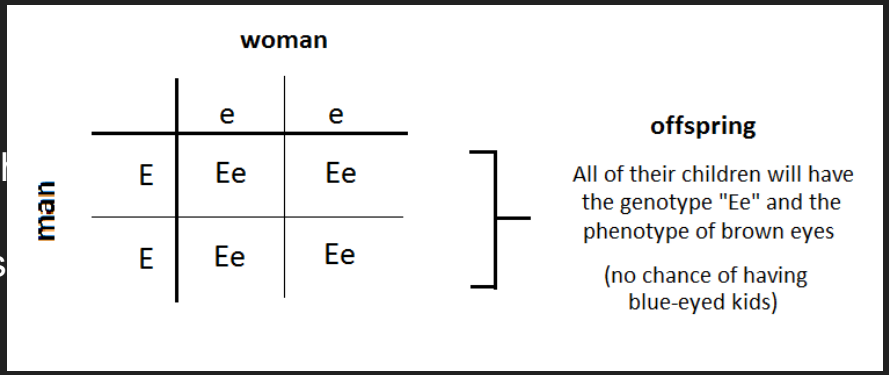


Punnett Squares

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 wit blue-eyed woman with the genotype ee ("homozygous recessive"). What is their chance of having a blue-eyed child?



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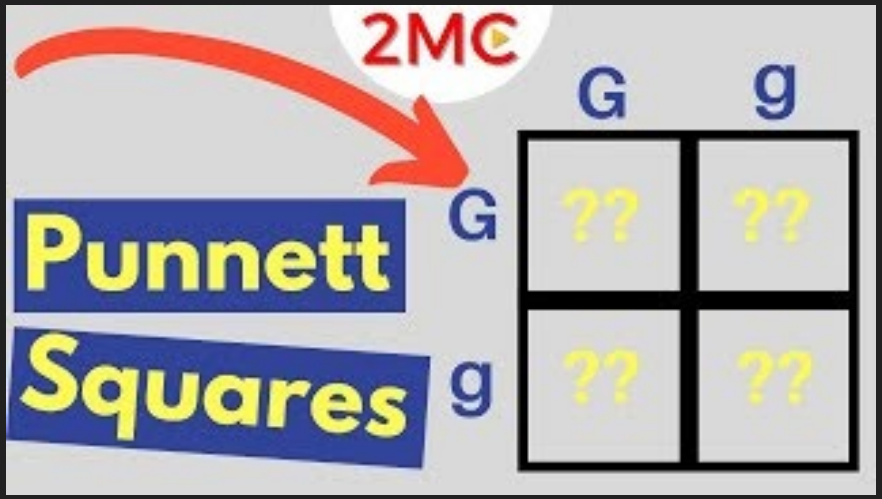
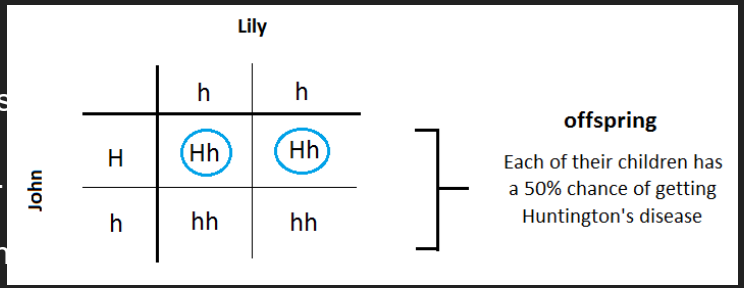


Genetic Counselling

An amniocentesis can be used to test a baby's genotype. A genetic counsellor also use Punnett Squares to predict whether the offspring of two individuals inherit a particular genetic disorder:

EXAMPLE:

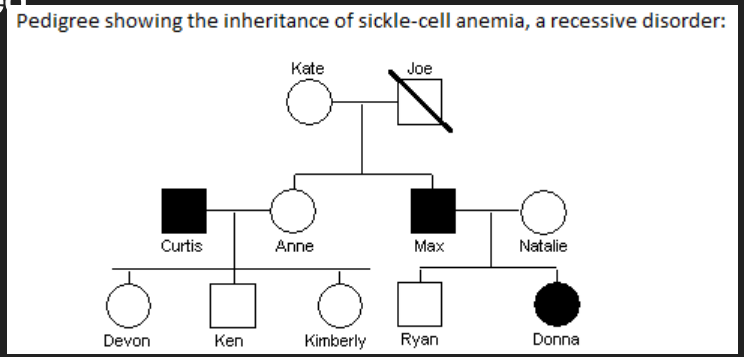
Huntington's disease is a dominant genetic disorder resulting in the degeneration of motor and cognitive abilities over time. John was diagnosed with Huntington's last year; with genetic testing, his genotype was determined to be Hh (heterozygous). John's wife, Lily, does not have Huntington's, so her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



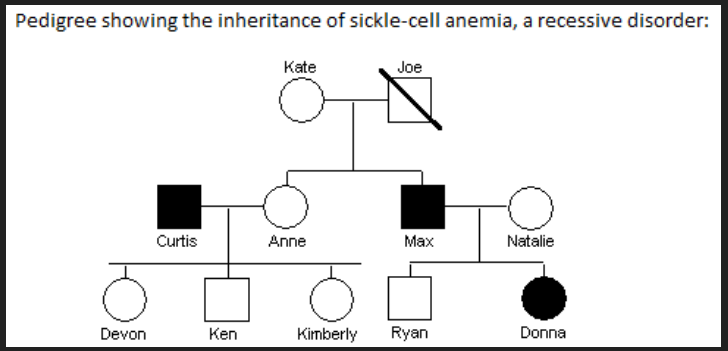
Pedigrees

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



Pedigrees



- 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 mou
- 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 accomplishment in the fie
genetics, it left many questions unanswered. Why are there so many seemingly useless gene
seemed as though the genetic code alone was not 100% responsible for making us who we a

- Epigenetics refers to how your behaviour and environment can affect how your genes are expressed
- Diet, exercise, the consumption of drugs & alcohol, and even mental health can attribu to the "flipping" of these switches
- Examples include thalidomide "flipper babies", fetal alcohol syndrome, and the effects of taking testosterone or estrogen

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

Epigenetics: An Indigenous Perspective

INDIGENOUS ELDERS OFTEN SAY THAT MEMORY IS IN THE BLOOD AND BONE, THAT OUR STORIES ARE PASSED NOT JUST VERBALLY BUT THROUGH A KIND OF GENETIC MEMORY

- Indigenous ways of knowing hold their conceptions that provide a mirror to our Western perspectives. One example is the Anishinaabe creation story referencing the ‘stringing together of the seeds of life’ that speaks to the genetic code

- The Indigenous concept of blood memory, that we carry the wisdom of our ancestors in our blood. This ties to the epigenetics we just discussed.

Genetic Engineering

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

- Genetic engineering is the process of taking a gene from one spe
  and inserting it into the DNA of another. This may cause the orga
  to produce more or less of a particular gene, or express an entire
  trait altogether.

- Genetic engineering has allowed us to grow crops in regions they
  wouldn't normally be able to grow; it also allows us to grow bigge
  healthier, more nutritious crops

- Genetic engineering has allowed us to produce large amounts of
  insulin for people with diabetes; it is also the basis of many other
  medical breakthroughs in terms of disease treatment and prevent

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 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 t characteristics of their offspring?
- Is it ethical to clone humans?
- If you had a fatal genetic disorder, would you want to know?

Ethical Concerns