

DNA – Four letters, millions of possibilities



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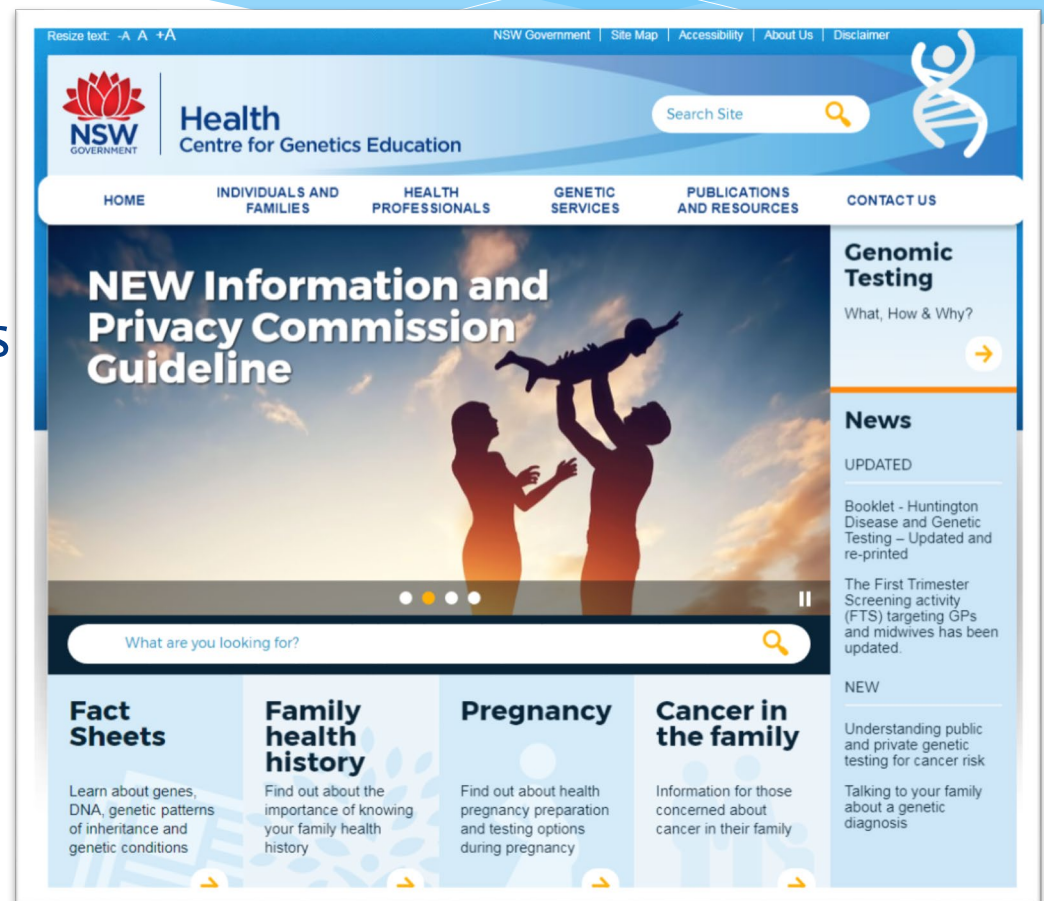
Overview

- * NSW Genetics Services
- * Genetics Basics
- * Why is genetic information “special”?
- * Ethical issues and scenarios



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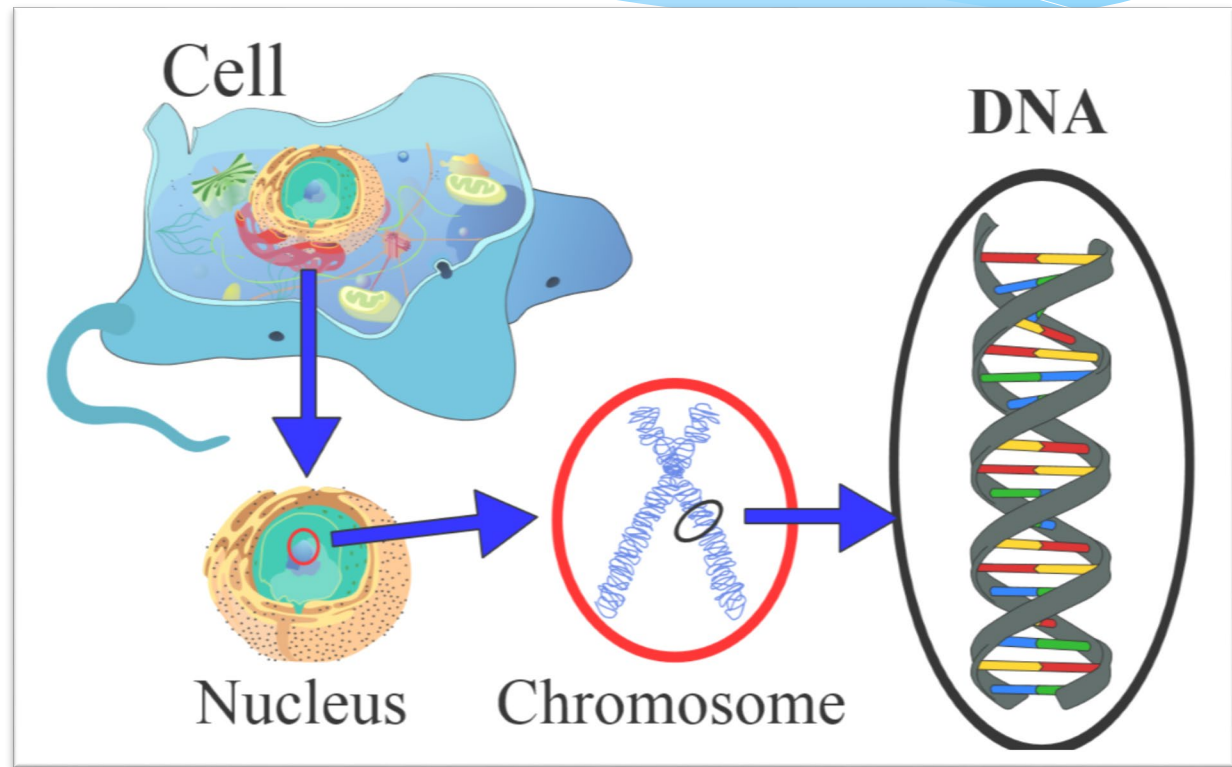
- * Website
- * Fact Sheets
- * Booklets and Brochures
- * Online Learning
- * List of Genetics Clinics
- * Contact



Genetics Revision



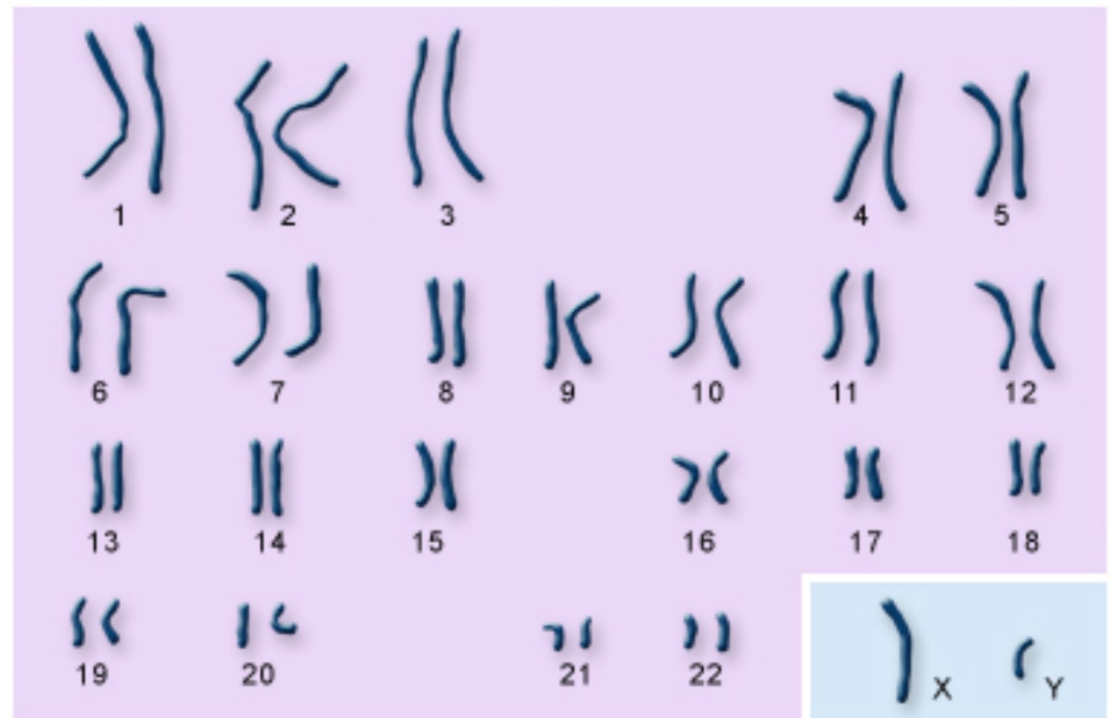
Genetics Revision



Chromosomes



Wilma 46, xx
Fred 46, XY



autosomes

sex chromosomes

U.S. National Library of Medicine

Genetics Revision

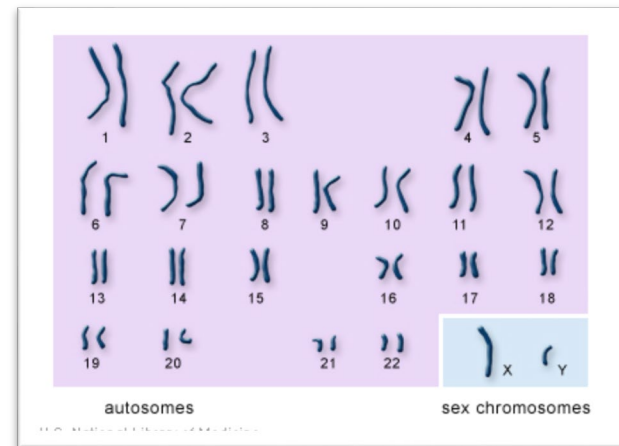


- * DNA
 - * 30 000 genes
 - * 4 letters
 - * Genes code for proteins which are the budding blocks of our body

Genetics Revision



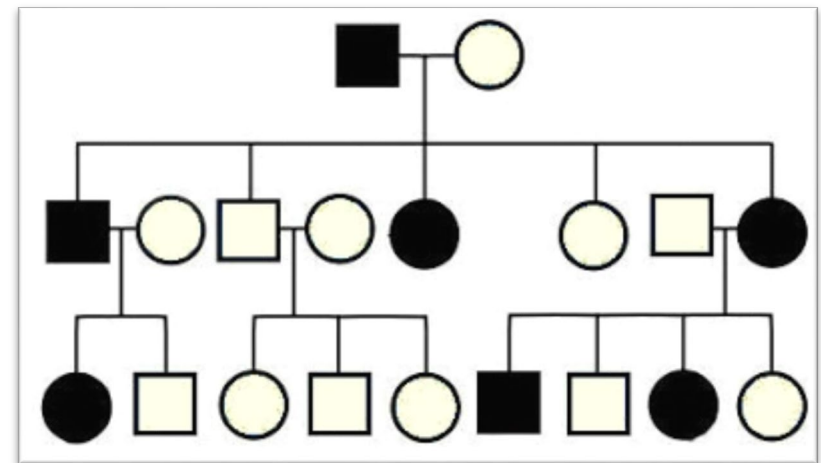
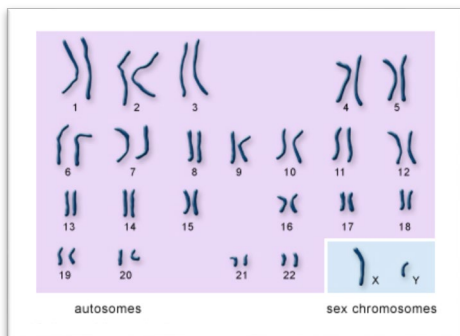
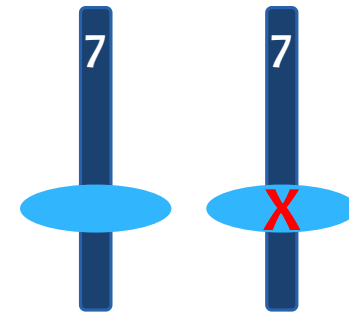
- * Genes, like chromosomes
 - * Come in pairs (body cells)
 - * Parents pass down one copy each so child has 2 of that gene
 - * Egg and sperm have one copy of each chromosome and therefore each gene



Genetic Inheritance

Autosomal Dominant

- * Autosomal Dominant
 - * Gene located on a numbered chromosome
 - * Appears in multiple generations
 - * One copy of gene mutation leads to symptoms
 - * Males and females affected
 - * 50% chance of passing gene on

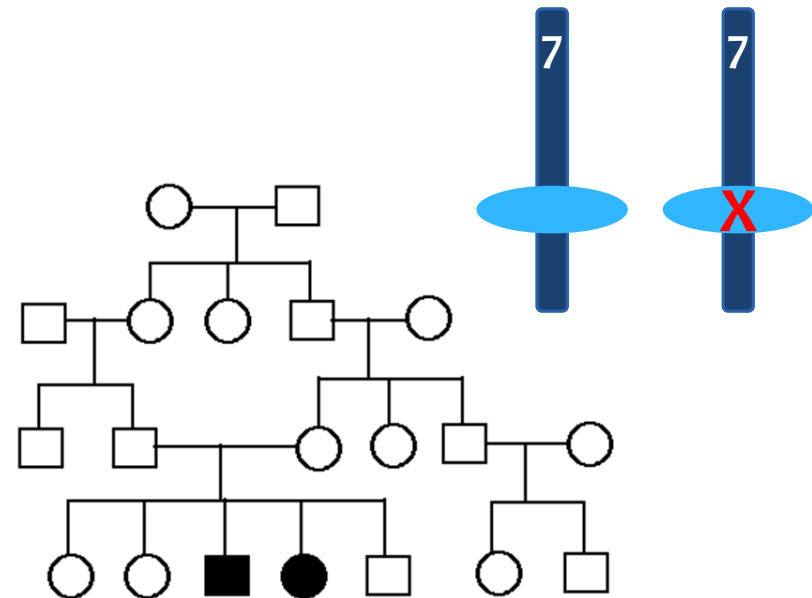
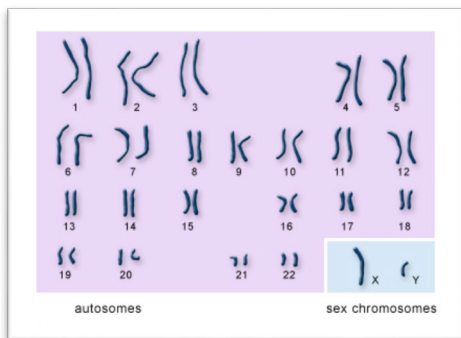


Genetic Inheritance

Autosomal Recessive

- * Autosomal Recessive

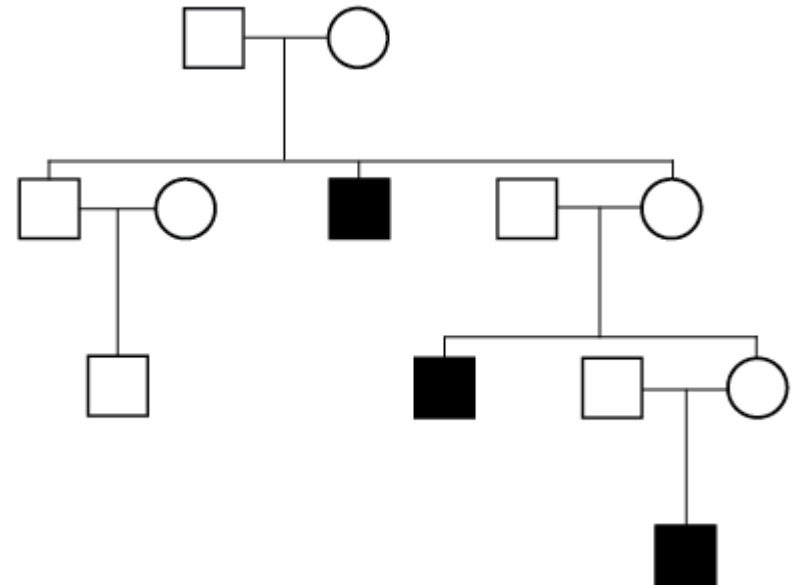
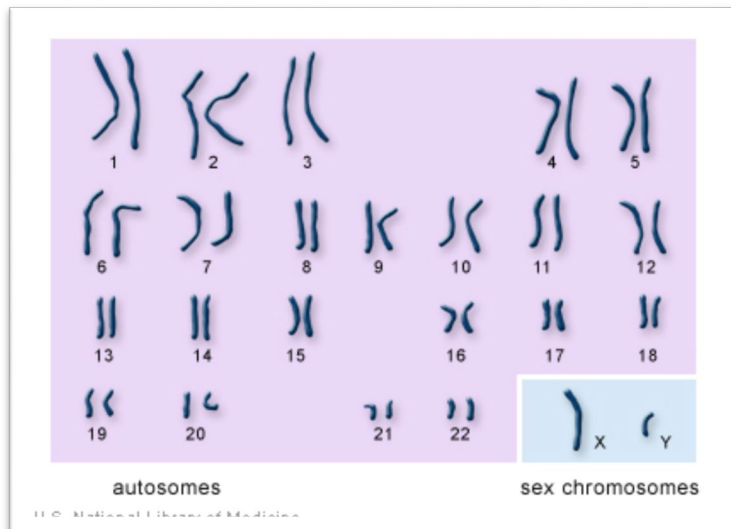
- * Gene located on a numbered chromosome
- * Appears in single generation
- * One copy of gene mutation makes you a healthy carrier
- * Two copies of gene mutation leads to symptoms
- * Males and females affected
- * 1 in 4 chance in each pregnancy



Genetic Inheritance

X Linked

- * X-Linked
 - * Gene located on X chromosome
 - * Males more affected than females (or no females)
 - * Males have no affected sons



Its Complicated!



Genetic Conditions

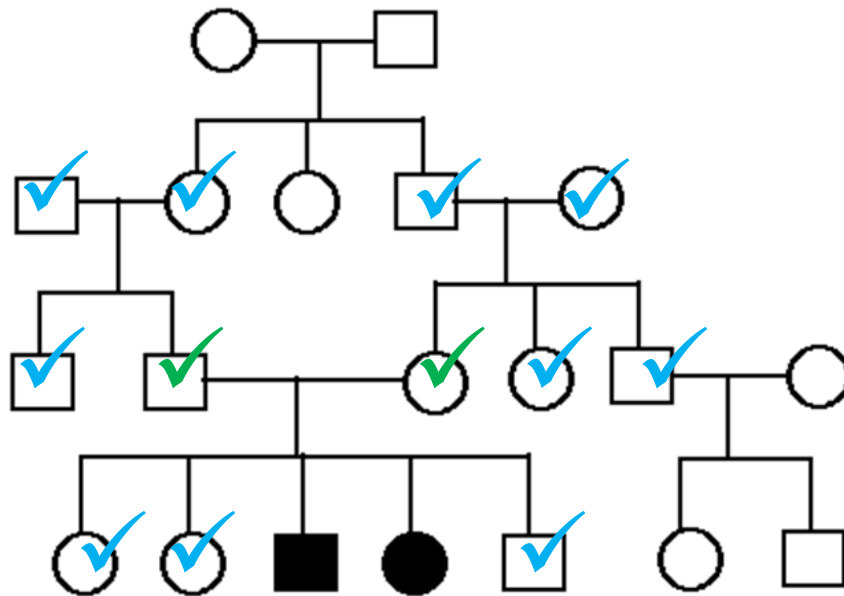
- * Accurate diagnosis of the person is important
 - * Accurate genetic risk
 - * Information for other family members
 - * Testing
- * Positive impacts
- * Negative impacts
- * Uncertainty



Ethics

- * Privacy - Who can access your genetic information?
- * Informed consent
- * What is “normal”?

Why is genetic information special?



✓ carrier

✓ possible carrier

