DNA – Four letters, millions of possibilities

Mona Saleh
Centre for Genetics Education
NSW Health
mona.saleh@health.nsw.gov.au
Overview

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

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

Wilma
46, xx

Fred
46, XY
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

* 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
X-Linked

- Gene located on X chromosome
- Males more affected than females (or no females)
- Males have no affected sons
It's Complicated!
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