Role Cards

1) Your DNA being tested

2) Sibling

3) Child

4) Partner

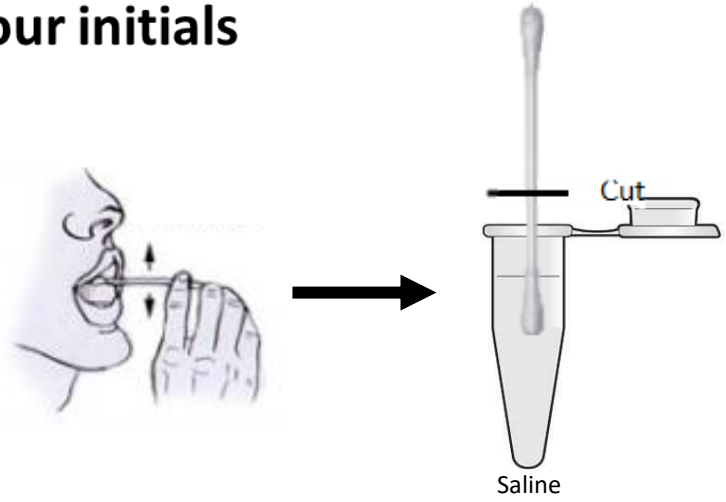
5) Friend

6) Parent

Activity 1:
DNA isolation – Mouth Swab

Mouth Swab

- Vigorously rub the inside of your cheeks (without hurting yourself!) using the cotton swab, for 30 seconds.
- Place the tip of the swab into the tube labelled ‘Saline’ and swirl, then press the swab against the walls of the tube to squeeze out any excess saline.
- Close the tube and mark it with your initials

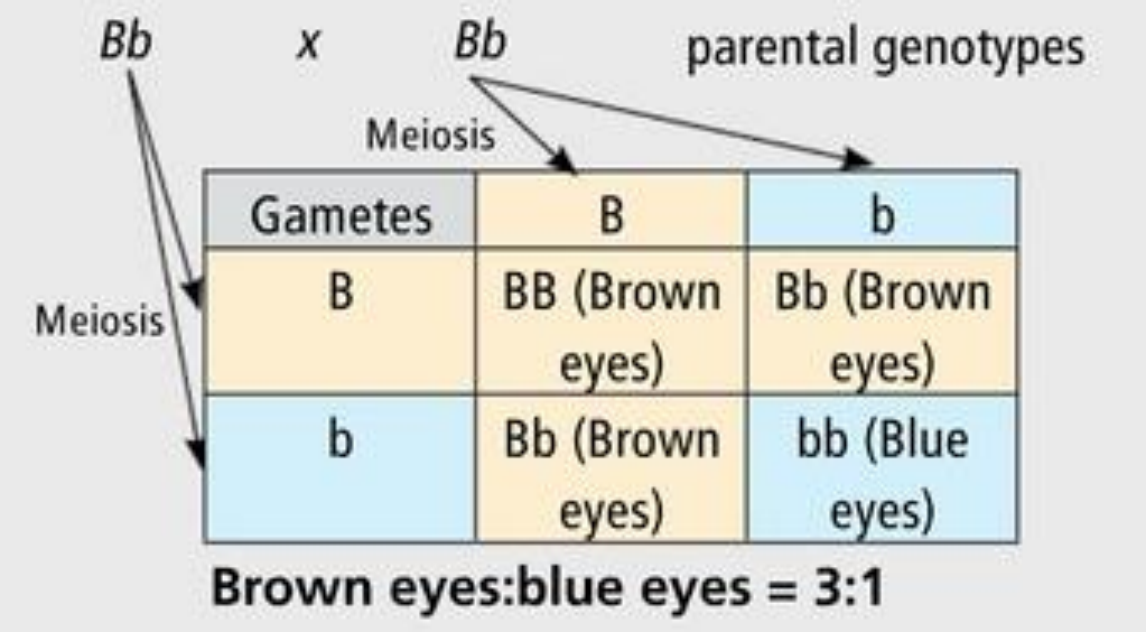


Activity 2: Genetic Crosses

Can you identify the genotype and phenotype of each individual?

- On your tables you will have a number of cards with letters on it representing the **genotype of two parents ('Bb'** and '**Bb')**
- In order to work out the genotypes and phenotypes you have '**B' x 4** and '**b' x 4** and **Brown Eyes x 4** and **Blue Eyes x 4**

Activity 2: Answers



Activity 3:
Step 3: Gel Electrophoresis

Load gel:

9 wells:

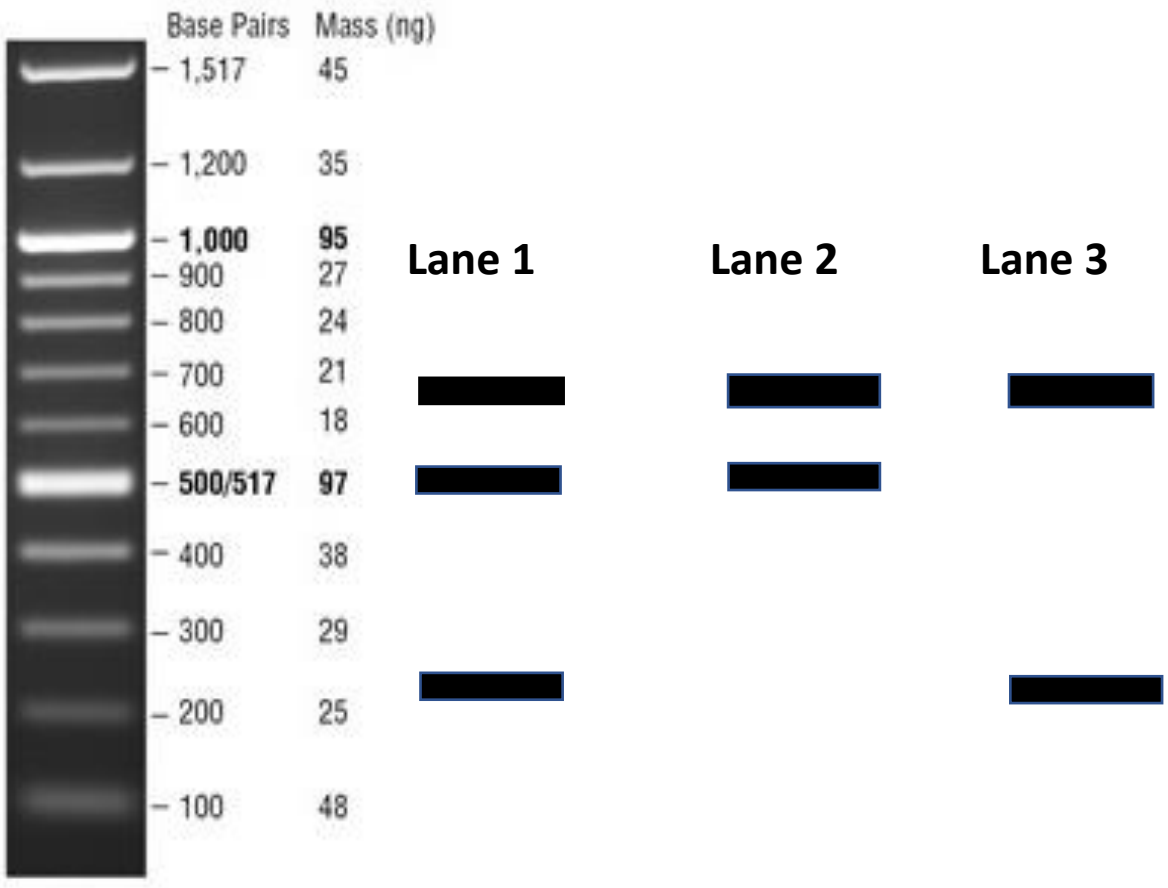
1	2	3	4	5	6	7	8	9
Sample: DNA ladder	A	B	C		A	B	C	
Volume: 10 µl	20 µl	20 µl	20 µl		20 µl	20 µl	20µl	

Don't forget to change the tip after each sample!

Suggested loading order

Activity 4: PTC gene

Can you identify the genotype and phenotype of each individual?

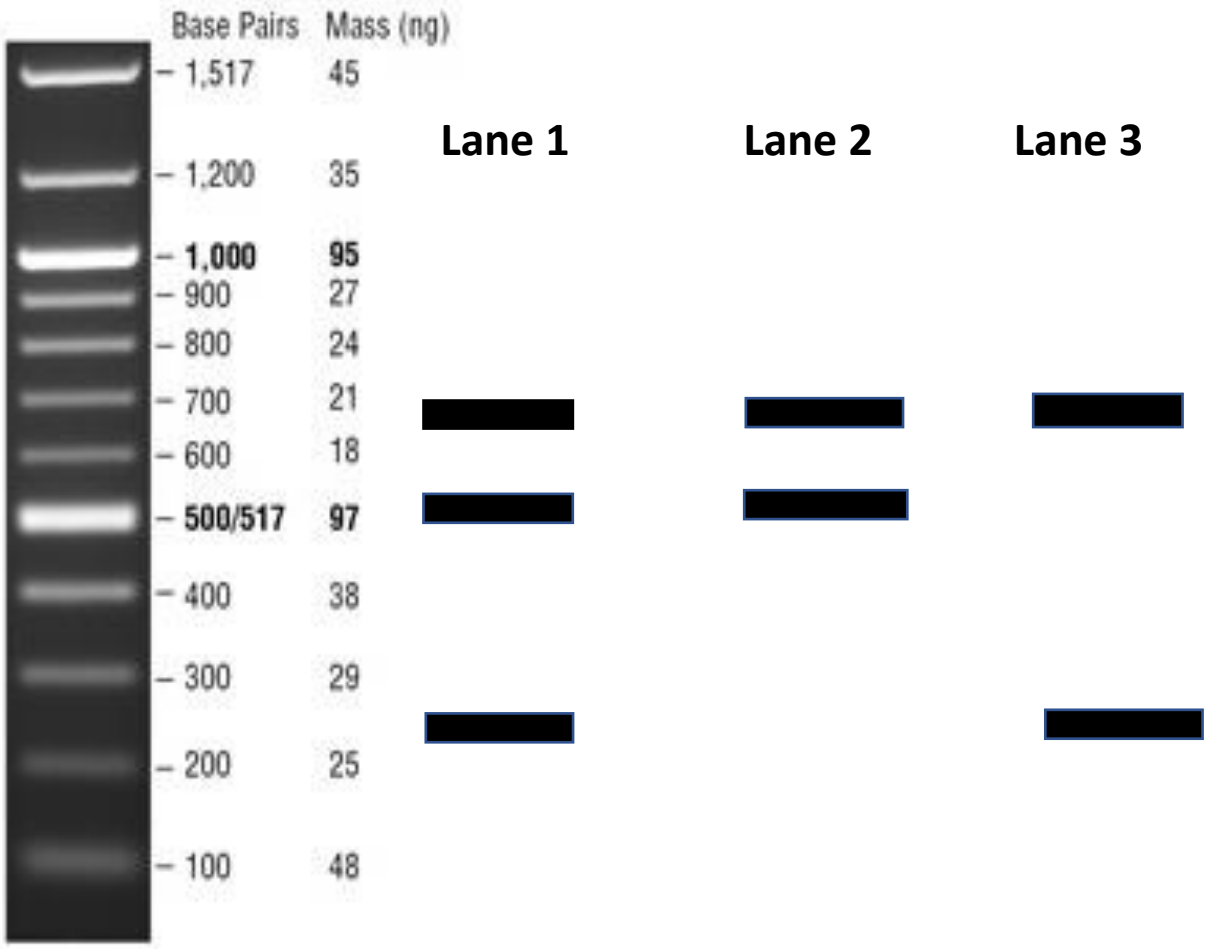


Control Band: 682 bp

t: 513 bp

T: 227 bp

Activity 4: Answers



What is the genotype and phenotype of each individual?

Lane 1: Tt – Heterozygous Taster

Lane 2: tt – Homozygous Non-Taster

Lane 3: TT – Homozygous Taster

Activity 5:
Sample Reports

You have bought the 23andMe kit, given your DNA sample, it has been amplified via PCR and here are your results

4 Sample Reports

Late-onset Alzheimer's Disease: Test revealed you **do not** have the e4 variant in the APOE gene tested for late-onset Alzheimer's Disease

Breast/Ovarian Cancer: Test detected you **do** have one variant (185delAG) in BRCA1 gene

Cystic Fibrosis: Test revealed you **do** have one variant (DeltaF508) in the CFTR gene

Tay-Sachs Disease: Test revealed you **do** have one variant (G269S) in the HEXA gene

Activity 5:
Sample Reports:
e4: Late-Onset Alzheimer's Disease

e4 – tests for variant in the APOE gene associated with Late-Onset Alzheimer's disease (a form of dementia)

Result: You **do not** have the e4 variant we tested associated with Late-Onset Alzheimer's disease

Report details:

- Does not include all variants or genes associated with late-onset Alzheimers disease
- Does not include any variants or genes linked to early-onset Alzheimer's disease
- Keep in mind that other factors influence your risk including age, sex, family history, heart health, diet and intellectual ability

Activity 5:
Sample Reports:
BRCA1: Breast/Ovarian Cancer

BRCA1/BRCA – genetic risk based on a limited set of variants for breast, ovarian and other cancers

Result: Female – one variant (185delAG) detected in BRCA1 gene

Report details:

- greatly increased risk of developing breast and ovarian cancer
- 45-85% of women with a BRCA1 variant develop Breast Cancer
- 39%-46% of women with BRCA1 variant develop Ovarian Cancer
- potentially higher risk of pancreatic cancer

Activity 5:
Sample Reports:
DeltaF508 – Cystic Fibrosis

DeltaF508 – carrier status test for variant associated with cystic fibrosis (CF – a progressive respiratory condition)

Result: one variant (DeltaF508) detected in CFTR gene

Report details:

- You are a carrier for CF
- You do not have CF
- Your biological children have an increased chance of having CF
- If your partner is also a carrier, each child has a 1 in 4 (25%) chance of having CF.

Activity 5:
Sample Reports:
G269S: Tay-Sachs Disease

G269S – carrier status test for variant associated with Tay-Sachs disease (progressive, fatal neurological disorder that begins in infancy)

Result: one variant (G269S) detected in the HEXA gene

Report details:

- You are a carrier for Tay-Sachs disease
- You do not have Tay-Sachs disease
- Your biological children have an increased chance of having Tay-Sachs disease
- If your partner is also a carrier, there is a 1 in 4 (25%) chance in each pregnancy that the child will be affected by Tay-Sachs disease

Activity 5:
23andMe: Personal Genome Service

'for the purpose of reporting and interpreting genetic health risks. It is not intended to diagnose any disease'
'Each genetic health risk report describes if a person has variants associated with a higher risk of developing a disease, but does not describe a person's overall risk of developing the disease.'

*Warnings & Limitations:

This report does not include variants in other genes linked to hereditary cancers and the absence of variants included in this report does not rule out the presence of other genetic variants that may impact cancer risk.'
Results should be confirmed in a clinical setting'