STEAM

IN JUNIOR CYCLE

AMGEN" Biotech Experience

Scientific Discovery for the Classroom

Ireland

Genetic Testing & Prediction of Disease

About Amgen Biotech Experience

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Advanced **M**olecular **Gen**etics (**AMGen**)

- **Unlocking the potential of biology for patients suffering from serious illnesses**
- **Focus on oncology/haematology, cardiovascular disease, inflammation, bone health, nephrology and neuroscience.**
- **Science education initiative began 25+ years ago in USA**
  - ✓ **Motivate students through hands-on experience**
  - ✓ **Use industry standard equipment**
  - ✓ **Linking the techniques to real-world examples**

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JCT Workshop Overview

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- Advances in science/technology have changed the way we can learn about ourselves.

- Can profile your own genetic information to learn not only about your ancestry but also about your health risks e.g. **23andMe genetic testing kit**

- Information held within our DNA can lead to many ethical/ societal issues that historically we did not have to consider.

- **This workshop will focus on:**
  a. societal implications of genetic testing
  b. practical aspects of the process of genetic testing

Health & Safety

Why are these precautions necessary?

- To protect yourself from harmful reagents
- To protect samples from contamination

When you are wearing gloves

- Don’t scratch your nose
- Don’t touch door handles
- Don’t answer your mobile phone
- Don’t touch food

If necessary, take one glove off

Genetic Testing - 23andMe

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Ancestry Service

Experience your ancestry in a new way! Get a breakdown of your global ancestry by percentages, connect with DNA relatives and more. learn more

€99

(add to cart)

RECOMMENDED

Health + Ancestry Service

Get an even more comprehensive understanding of your genetics. Receive 90 online reports *on* your ancestry, traits and health - and more. **New BRCA1 / BRCA2** (Selected **Variants)*** **report just added!** learn more

€169

add to cart

Important Test Info

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Activity 1:
DNA isolation – Mouth Swab

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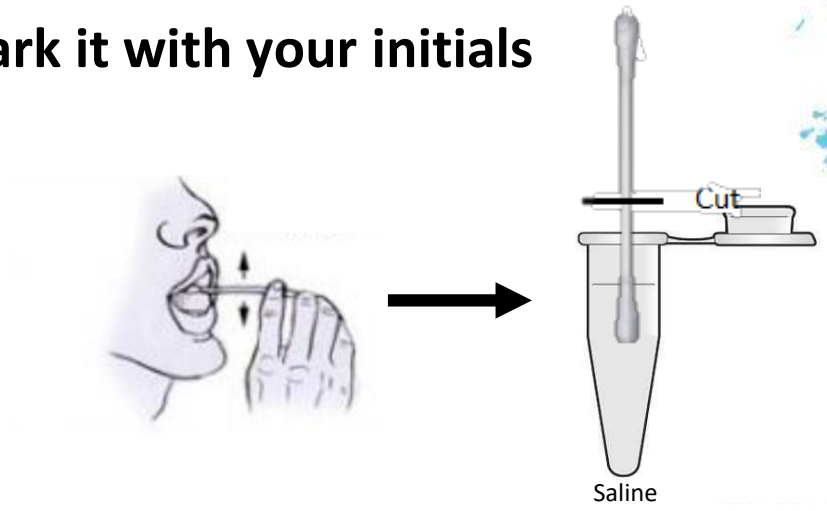
Mouth Swab

- **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**



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Step 1: DNA isolation - Overview

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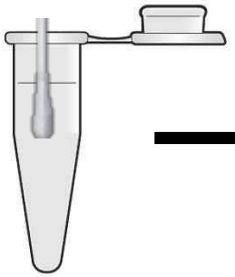
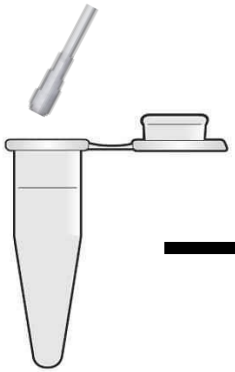
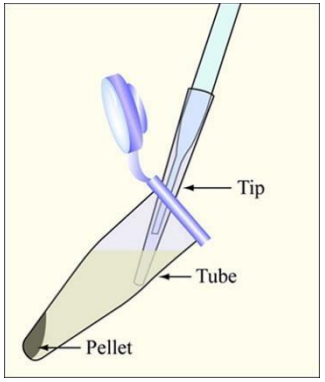


Saline + Swab tip

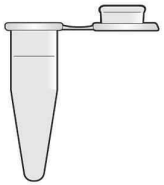


Remove swab

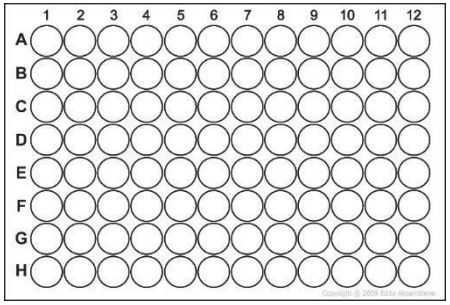
Micro-Centrifuge
90s



Remove Supernatant
(P1000)
Resuspend cells (P20)



Transfer 20ul to
Chelex in (small)
PCR tube



Place in PCR machine

Micro-Centrifuge
90s

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Step 2: Polymerase Chain Reaction (PCR) - Overview

Video Available at:
https://youtu.be/2KoLnIwoZKU

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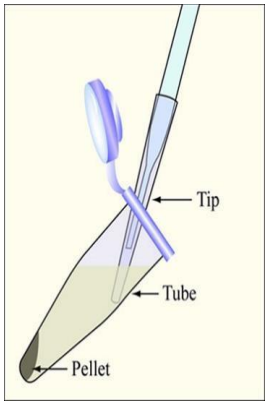
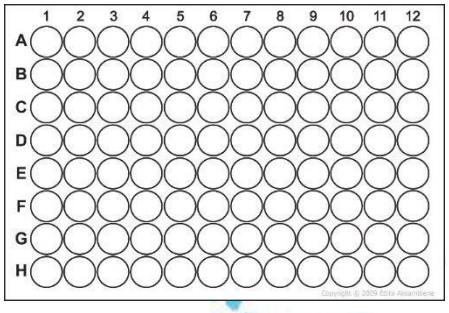
Step 2: PCR - Workflow

Micro-Centrifuge 90s



Remove 5 µl of the clear supernatant

Transfer the clear supernatant into another PCR tube containing PCR primer, Polymerase, dNTPs and Mg ions.



Place in PCR machine

Add DNA loading dye to the PCR samples

Load the Samples on 2% agarose gel along with a DNA ladder.

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Patterns of Genetic Inheritance

- **Variant:** an alteration in the most common DNA nucleotide sequence
- **Allele**: different versions of the same variant are called alleles
- **Homozygous**: two alleles are the same
- **Heterozygous**: two alleles are different
- **Dominance**: one allele masks the effects of another allele
- **Recessive**: allele's effect is only expressed in the homozygous condition

Activity 2: Genetic Crosses

Can you identify the genotype and phenotype of each individual?

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- 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

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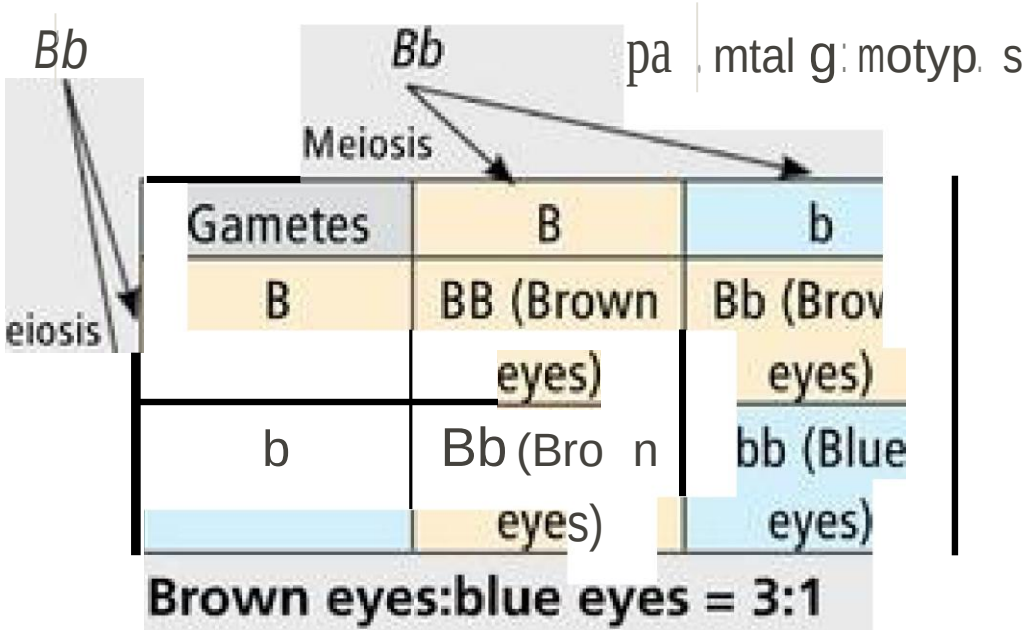


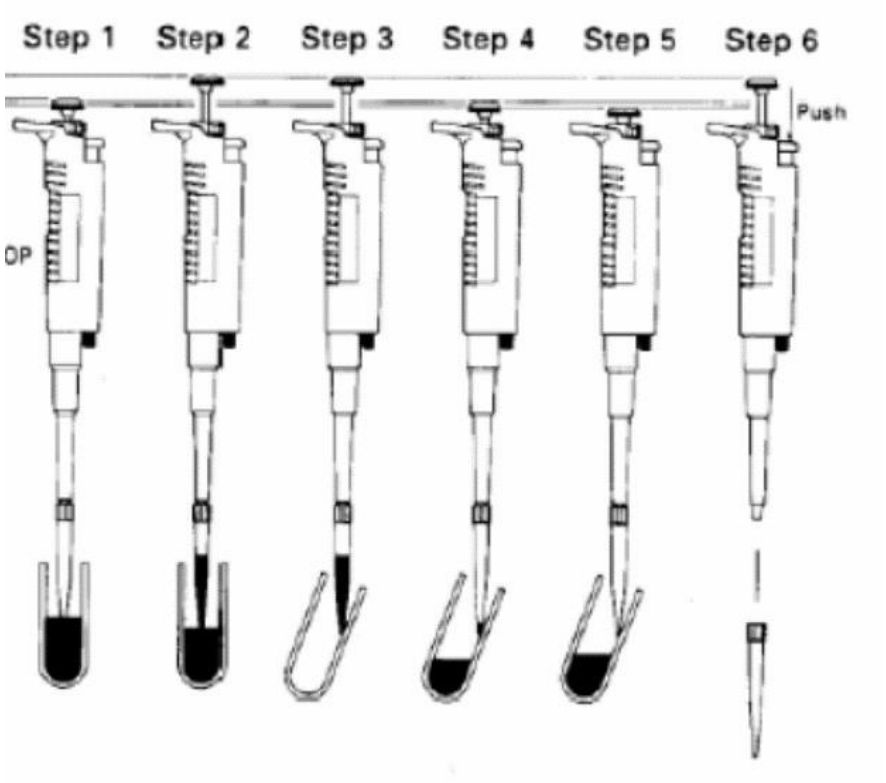
Bitter Taste Perception

- Testing for a single genetic traits - **bitter taste**
- Needed to detect rancid or poisonous food
- **Phenlythiocarbamide (PTC)**
  - Tastes very bitter or is virtually tasteless.
  - Taste sensation depends on genetic makeup of the taster
- TAS2R38 is the gene for the PTC taste receptor
  - Genetic sequence of TAS2R38 varies within the human population

❼Identification of Single Nucleotide Polymorphism (SNP) by PCR

-> Differentiate between the genetic profiles of a 'Taster' and a 'non-Taster'

Pipetting with a micropipette



1st stop **1st stop** **2nd stop**

Pressing plunger down to the 2nd stop will remove the last drop from the tip

Practice: Transferring 20µl liquid from one tube to another

Pipetting precautions

When loading the micropipette, **only press the plunger to the first stop** or you will draw too much solution into the pipette tip.

When drawing up the solution**, move your thumb gently upwards** – if you let go too fast, you will end up with the wrong amount and drops in the upper part of the tip.

Do not lay down a micropipette with fluid in the tip or hold it with the tip pointed upward because fluid could leak back into the pipette.

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

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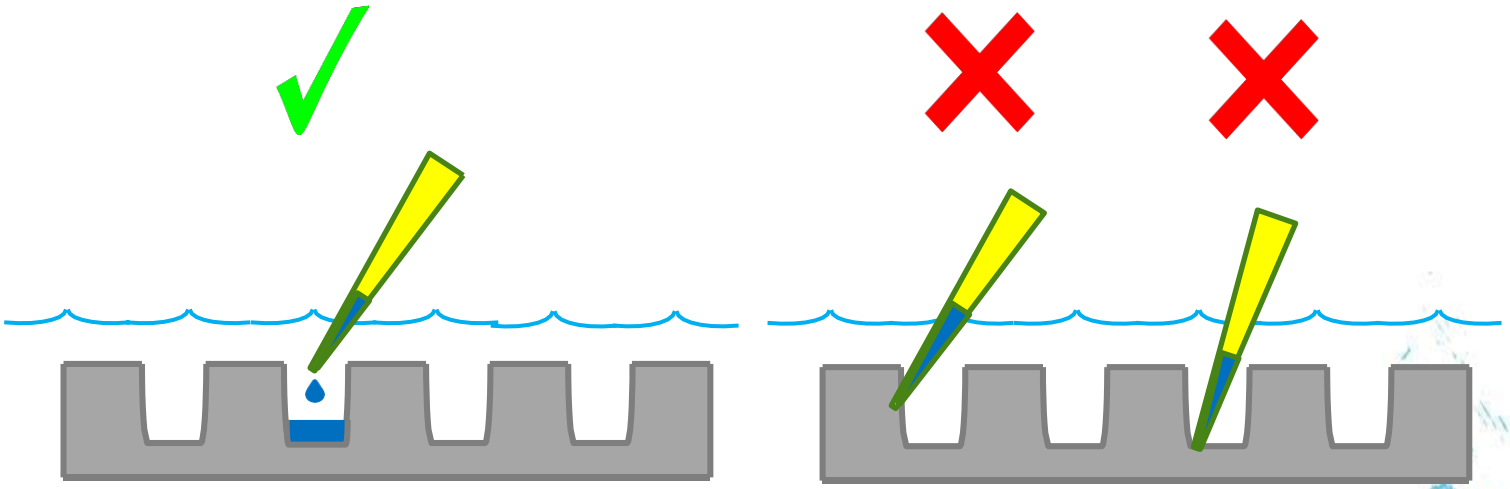
Gel Electrophoresis:

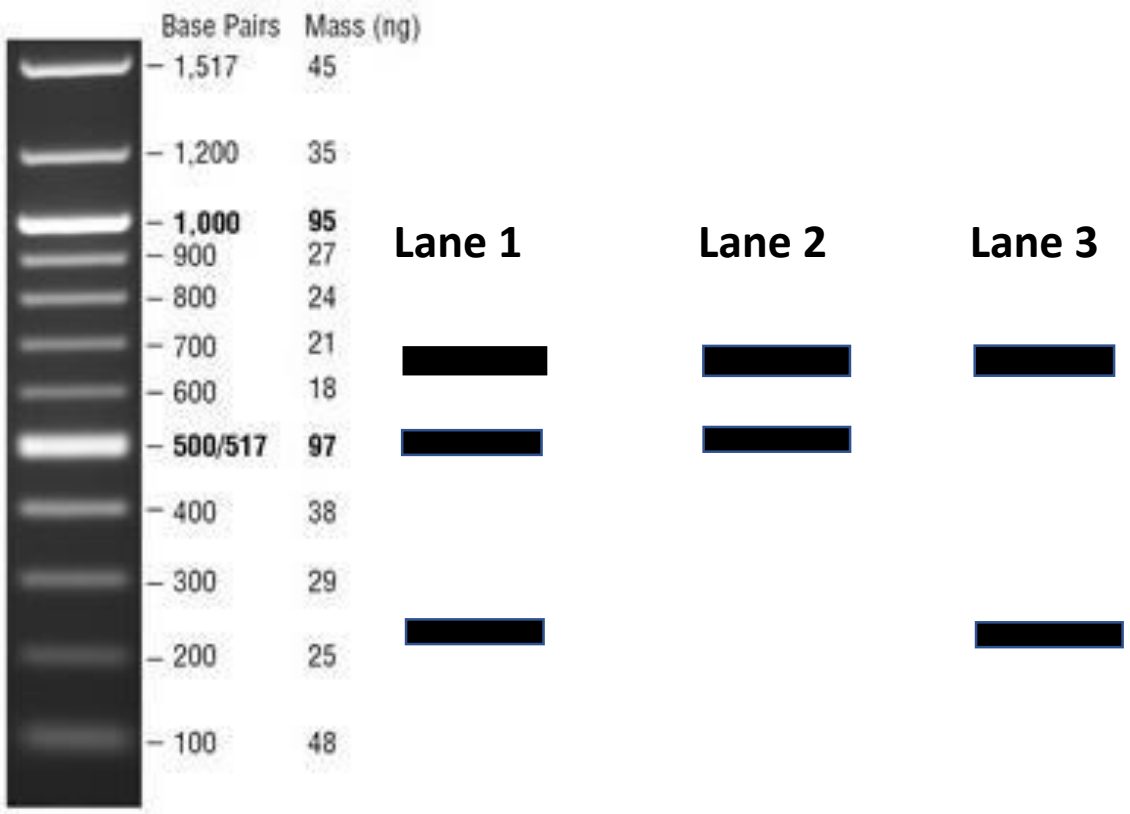
Precautions for loading the gel

Try not to jab through the bottom or side of the wells.



Activity 4: PTC gene

Can you identify the genotype and phenotype of each individual?



Control Band:

682 bp

t: 513 bp

T: 227 bp

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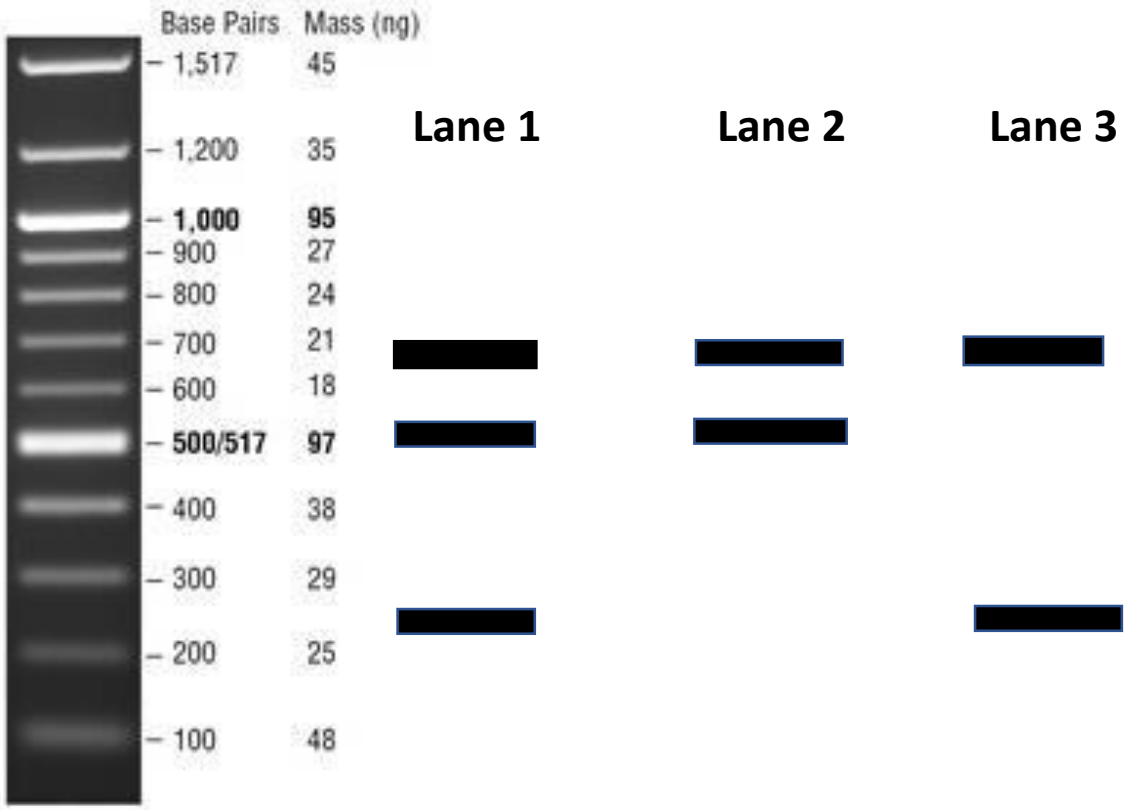
Activity 4: Answers



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

PTC PCR Results

tt – homozygous non-taster

DNA ladder

Tt – heterozygous taster

TT – homozygous taster

- Largest band: control band, allele independent
- Intermediate band: Non-functional allele (t)
- Shortest band: Functional allele (T)

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23andMe Reports
‘Live in the Know’

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Genetic Health Reports – tells you about genetic variants associated with increased risk for certain health conditions

Examples:

- **BRCA1/BRCA** – genetic risk based on a limited set of variants for breast, ovarian and other cancers
- **Late-Onset Alzheimer’s Disease** – genetic risk for a form of dementia

23andMe Reports
'Live in the Know'

Carrier Status Tests – detect genetic variants that that can cause inherited conditions

Examples:

- **Tay-Sachs Disease** – progressive, fatal neurological disorder that begins in infancy
- **Cystic Fibrosis** – genetic disorder affecting the lungs (but also pancreas, liver, kidneys and intestine)

Sample Reports

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Late-Onset Alzheimer•s Disease

Alzhe im er 's d isease is cha racte r ize d by memory loss, cogn itive d ecli ne, and pe rso na lit y changes. Late -onset Alzhe im e r's d iseas e i s the most common form of Alzhe im e r's d ise ase , d ev elo ping aft er age 65. Many facto rs, i n clud ing genetics, can infl uence a p erson 's chances of d eve lop ing t he cond ition. This t es t in cludes the most common genetic variant assoc i ated with late -o nset A lzheim er's d isease .

Jamie, you do not have the e4 variant we tested.

Your risk fo r Alzhe ime r' s disea se also d epen ds o n ot her fact ors, inclu d ing lifesty le, enviro nment, and gene t ic vari ants not cove red by th is t est.

0 variants detected

in the APOE gene

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Activity 5:
Sample Reports

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Bought the 23andMe kit, given your DNA, DNA has been amplified via PCR and here are your results - Discuss

4 Sample Reports

- Late-onset Alzheimer's Disease
- Breast/Ovarian Cancer
- Cystic Fibrosis
- Tay-Sachs Disease

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Are you willing to ‘live in the know’?

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THANK YOU!!

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PTC PCR Results

tt – homozygous non-taster

DNA ladder

Tt – heterozygous taster

TT – homozygous taster

- Largest band: control band, allele independent
- Intermediate band: Non-functional allele (t)
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