

# Human Polymorphisms

PTC Tasting

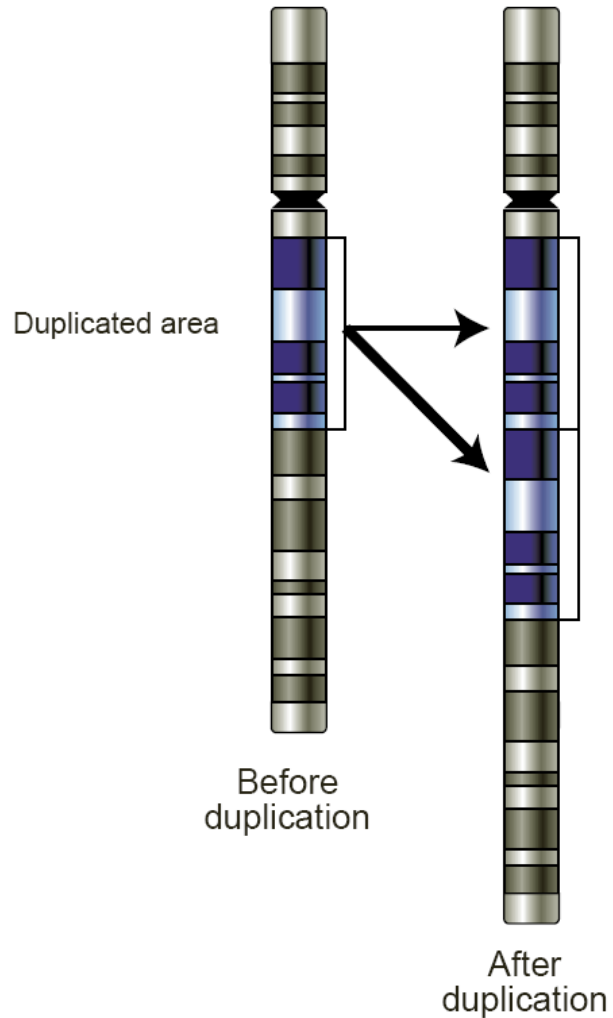
# Variation



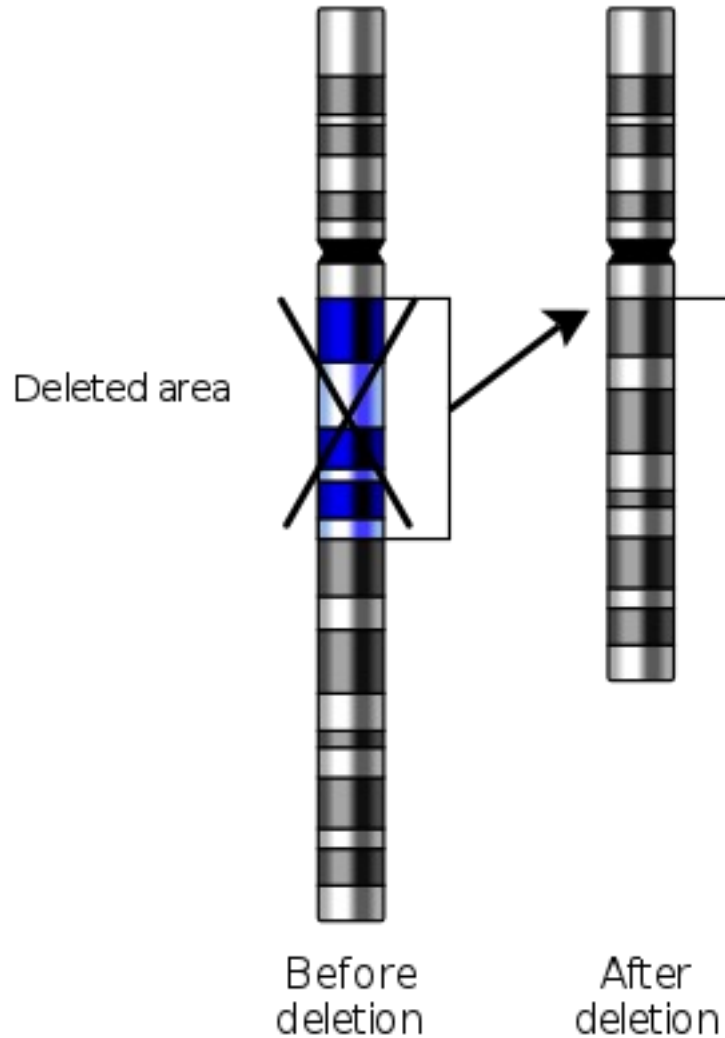
# Genetic Variations

## Copy Number Variation

- Different number of copies of the same gene



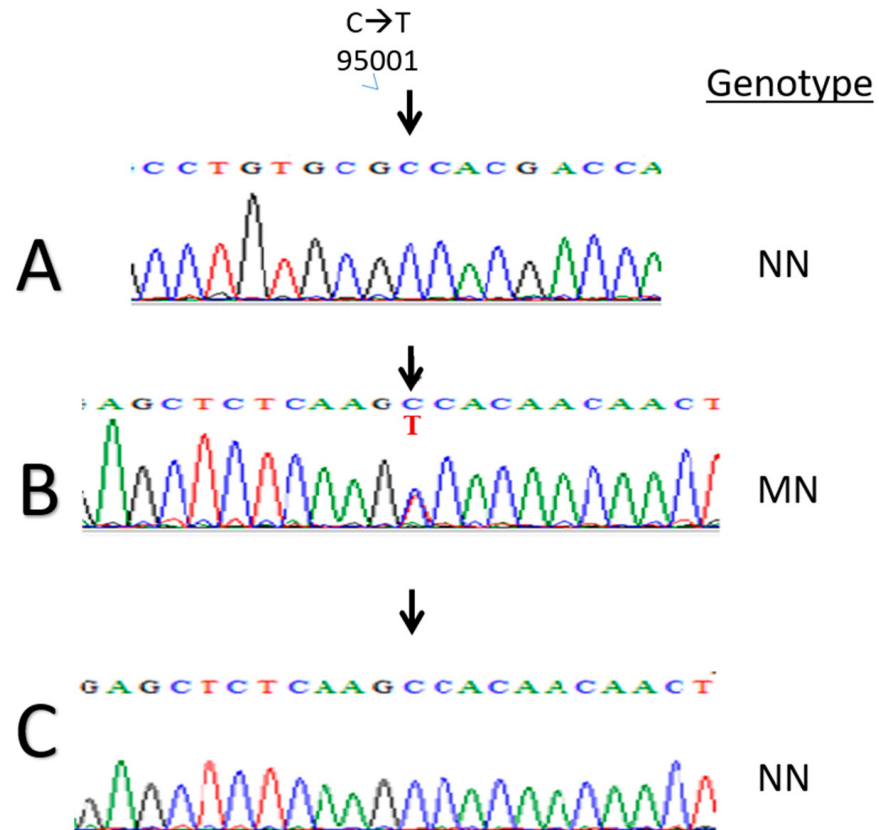
# Genetic Variations



## Copy Number Variation

- Genes may even get deleted

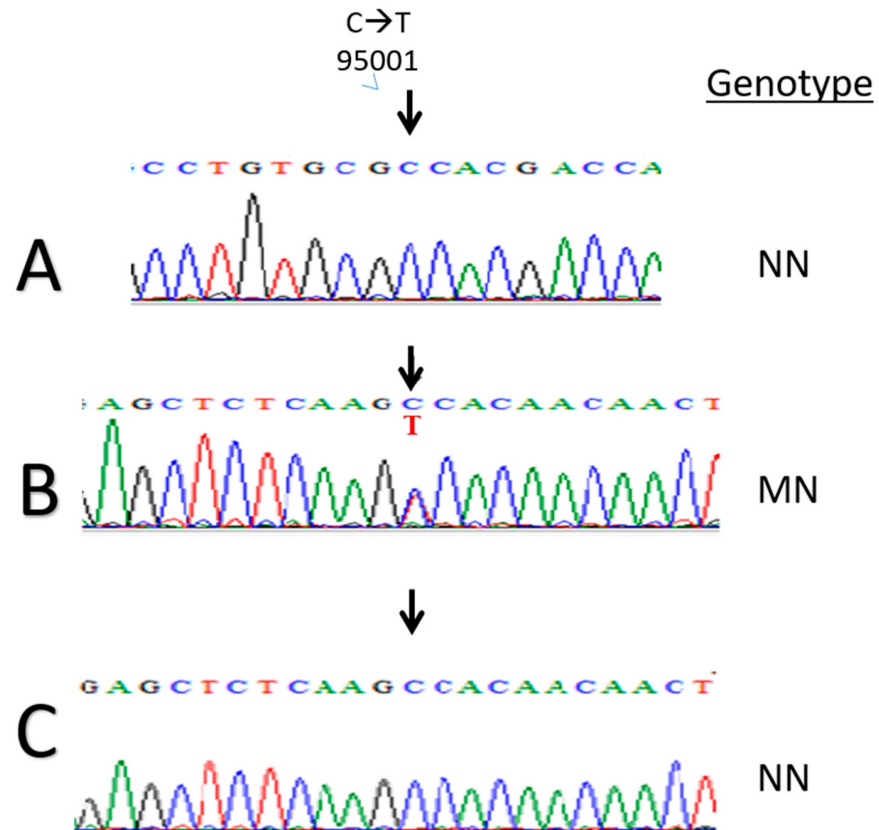
# Genetic Variations



## Polymorphism

- The gene may come in different varieties (poly- many/morphisim-form)
- When a single nucleotide has changed, we often call it a **SNP** (single nucleotide polymorphism)

# Genetic Variations



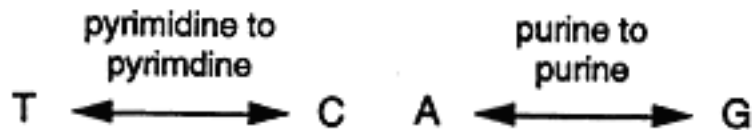
## Polymorphism

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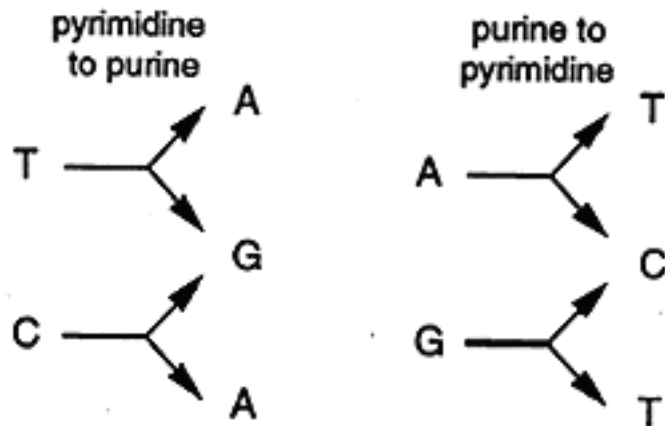
# SNP Classifications

## Transitions and Transversions

Transitions:



Transversions:

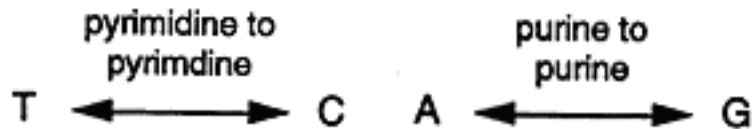


- Transition (more common)
  - a nucleotide is changed to another nucleotide with a similar chemical structure
- Transversion (less common)
  - a nucleotide is changed to another nucleotide that does not have the same chemical structure

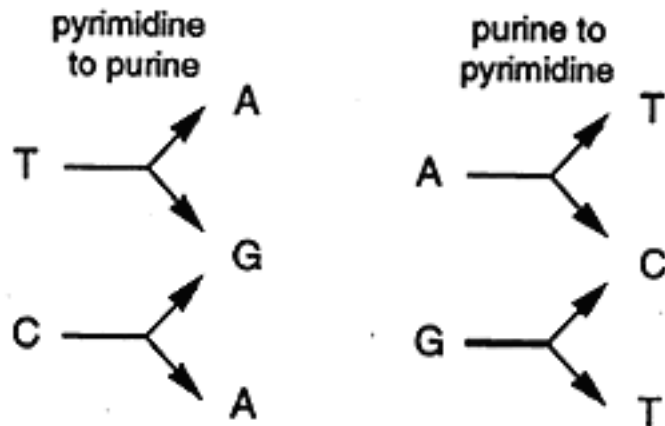
# SNP Classifications

## Transitions and Transversions

Transitions:



Transversions:








- Cytosine, Thymine – (pyrimidines)

- Adenine, Guanine – (purine)



# Most variations are subtle (phonological)



<b>British &amp; American English</b> 		
<a href="http://americanenglish.state.gov">americanenglish.state.gov</a>		
<b>British</b> chips		<b>American</b> french fries <i>/ˈfrɛntʃ ˈfraɪz/</i>
<b>British</b> crisps		<b>American</b> potato chips <i>/pəˈteɪtoʊ ˈtʃɪps/</i>
<b>British</b> biscuit		<b>American</b> cookie <i>/ˈkʊki/</i>
<b>British</b> peckish		<b>American</b> hungry <i>/ˈhʌŋɡri/</i>

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**A E** American English at State [americanenglish.state.gov](http://americanenglish.state.gov)

# Most variations are subtle (spelling)



## British & American English

Spelling Differences

americanenglish.state.gov

British		American
kerb		curb
plough		plow
mould		mold
liquorice		licorice
cosy		cozy

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**A E** American English at State

americanenglish.state.gov

ABCDEFGHIJKLMNOPQRSTUVWXYZ

# PTC Tasting Experiment

# Genetics of PTC Tasting

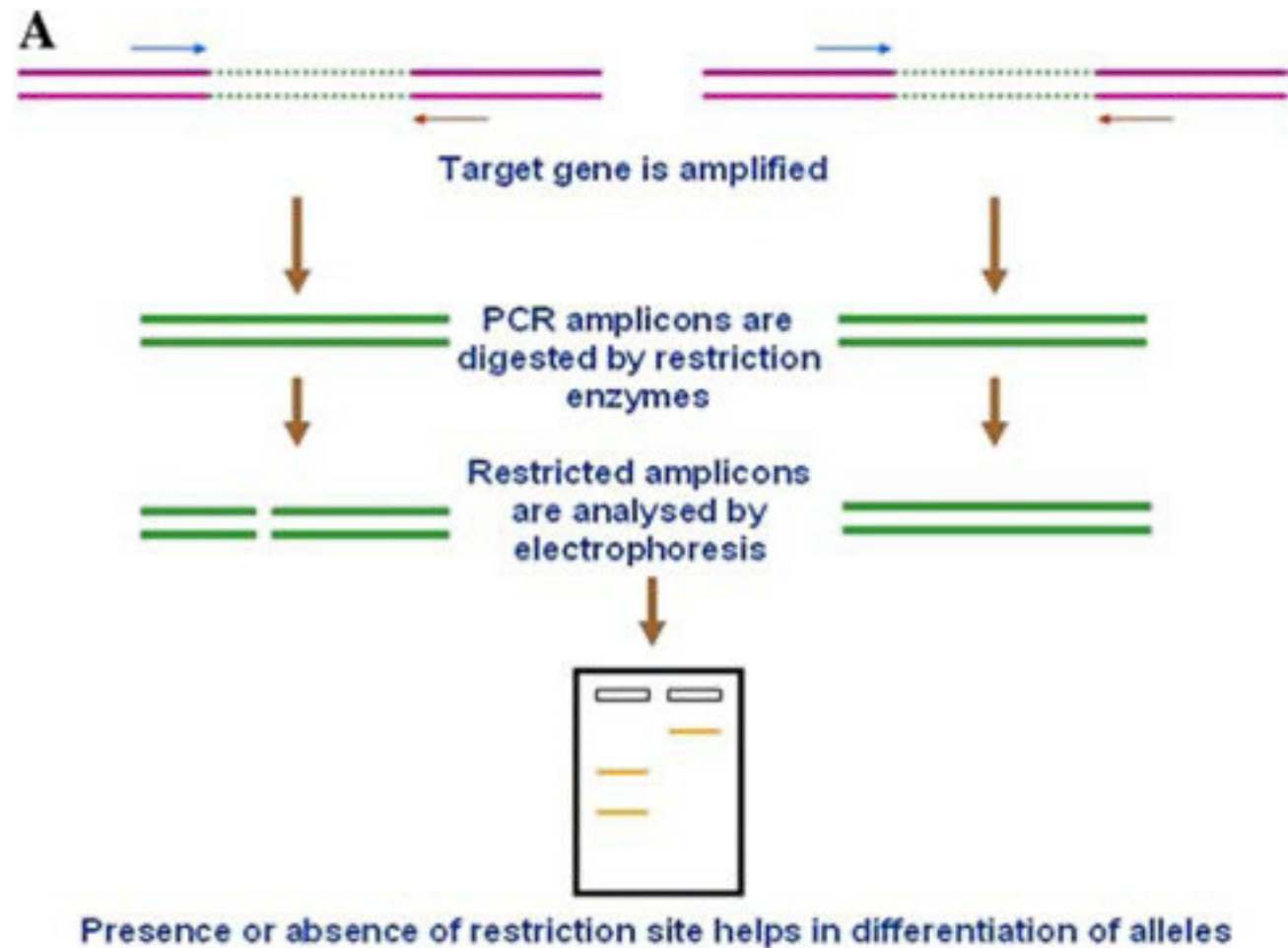
- PTC = phenylthiocarbamide
- Prepared by Arthur Fox at Du Pont Company in late 1920s
- Lab partner C.R. Noller complained of bitter taste
- Fox had no taste
- Followed up by Albert Blakeslee at Carnegie Department of Genetics
- Published in 1932



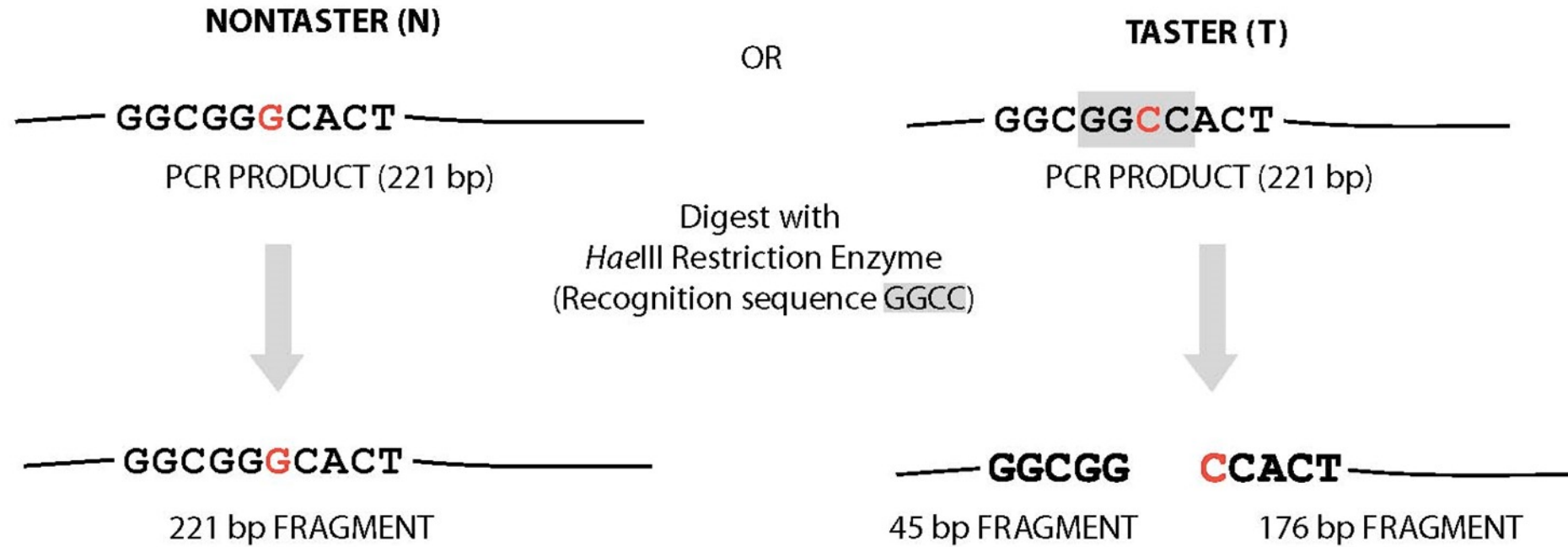
# Molecular Genetics of PTC Tasting

- Gene identified in 2003 by Dennis Drayna
- Polymorphism associated with PTC tasting
- Nucleotide position 145, (amino acid 49)  
Taster = C (proline) > Nontaster = G (alanine)

# CAPs (Cleaved, Amplified, Polymorphic Sequence)



## TAS2R Bitter Taste Receptor



# More Complication: PTC Haplotypes

Postition	Taster	Nontaster
145	C (proline)	G (alanine)
785	C (alanine)	T (valine)
886	G (valine)	A (isoleucine)