

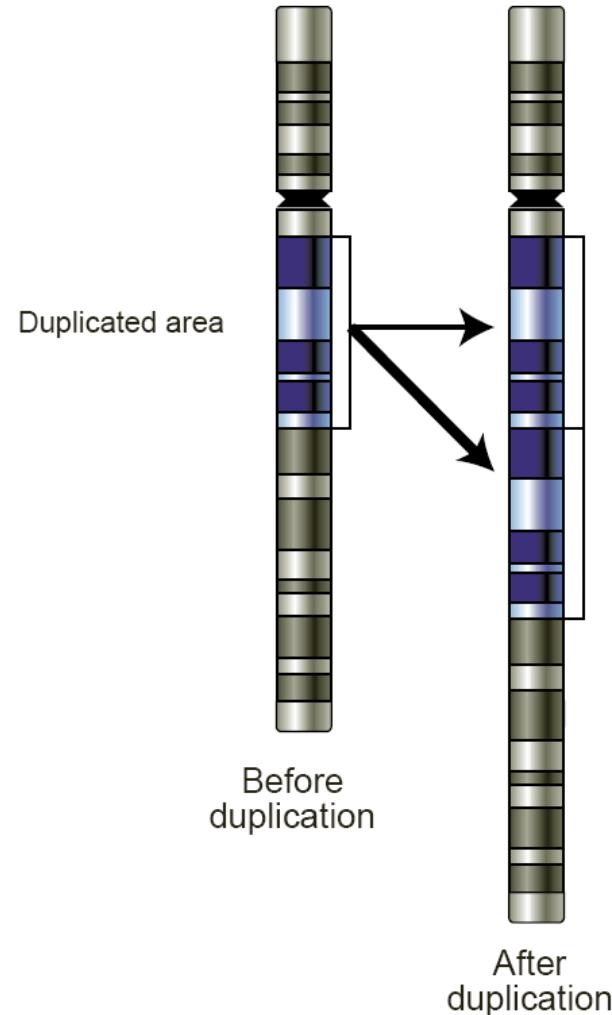
# Human Polymorphisms

PTC Tasting

# Variation



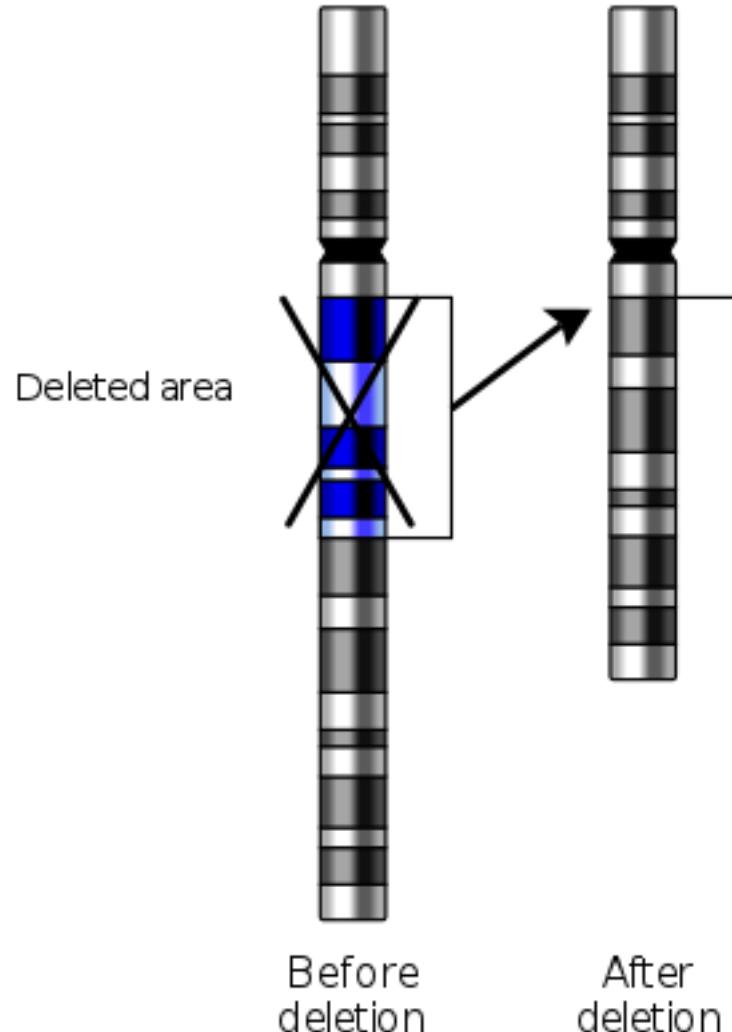
# Genetic Variations



## Copy Number Variation

- Different number of copies of the same gene

# Genetic Variations

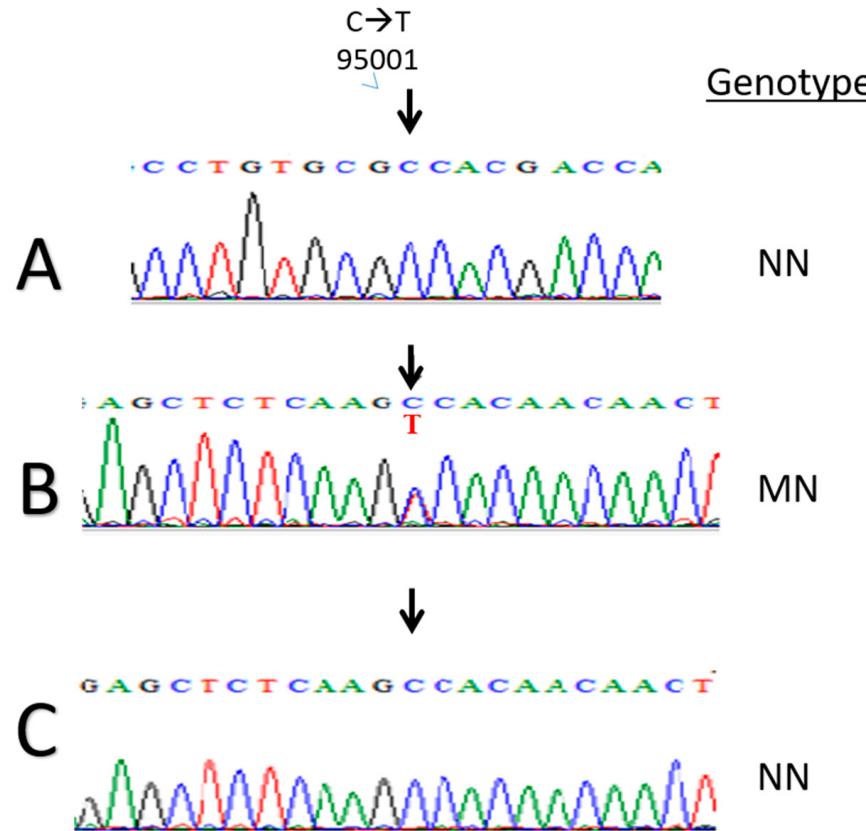


## Copy Number Variation

- Genes may even get deleted

The original uploader was [Mirmillon](#) at [French Wikipedia](#). - Vectorized from [File:Deletion.gif](#) National Human Genome Research [USA](#) Originally from [fr.wikipedia](#); description page is/was [here](#).  
Delección cromosómica.

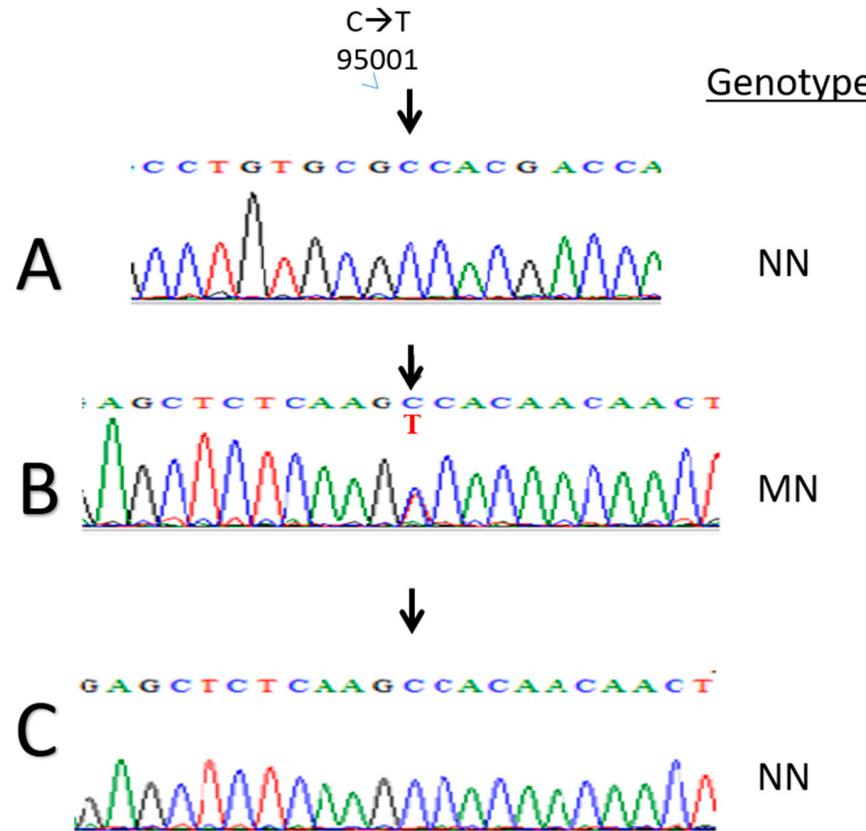
# Genetic Variations



## 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

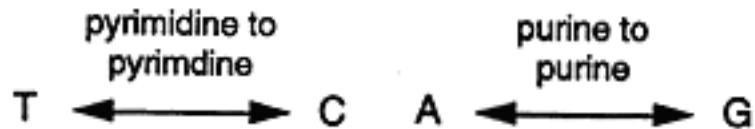


## Polymorphism

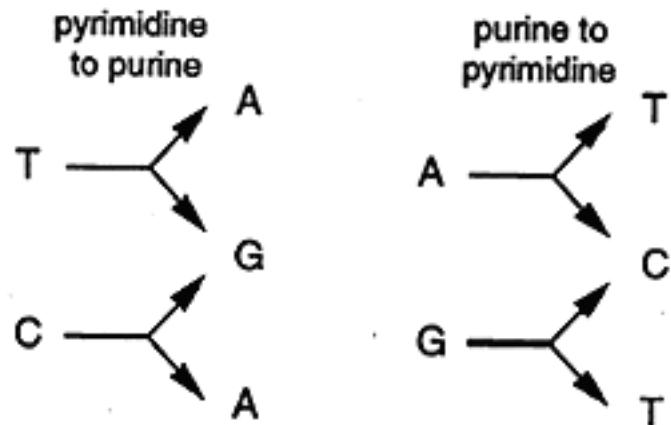
- The gene may come in different varieties (poly- many/morphisim-form)
- When a single nucleotide has changed, we often call it a **SNP** (single nucleotide polymorphism)

# SNP Classifications

Transitions:



Transversions:

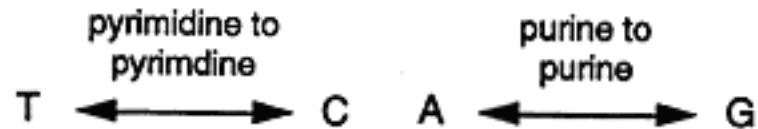


## Transitions and Transversions

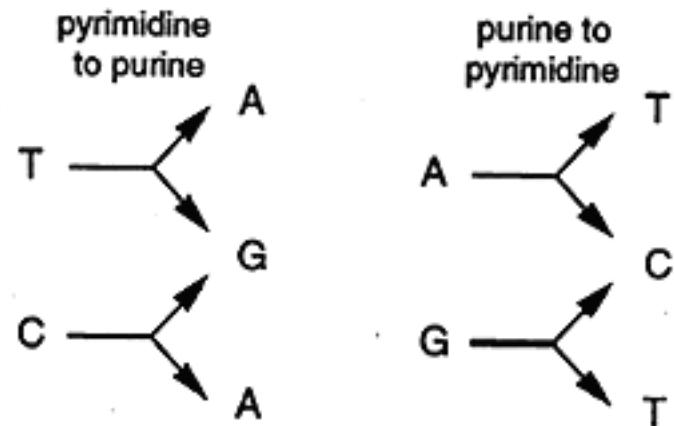
- Transition (more common)
  - a nucleotide is changed to another nucleotide with a similar chemical structure
- Transversion (less common)
  - a nucleotide is changed to another nucleotide that does not have the same chemical structure

# SNP Classifications

Transitions:



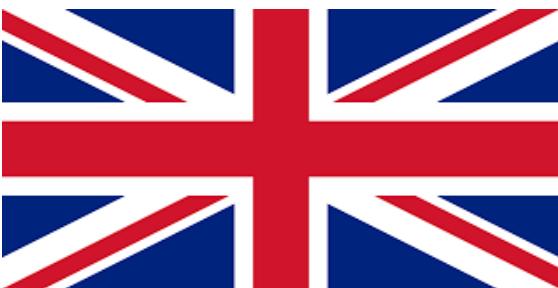
Transversions:



## Transitions and Transversions

- Cytosine, Thymine – (pyrimidines)
- Adenine, Guanine – (purine)

# Most variations are subtle (phonological)



**British & American English** 

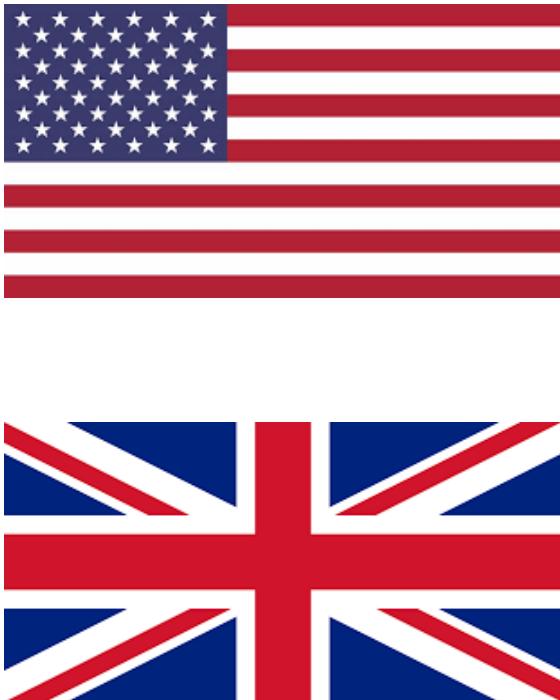
[americanenglish.state.gov](http://americanenglish.state.gov)

<b>British</b> chips		<b>American</b> french fries <i>/'frɛntʃ 'fraɪz/</i>
<b>British</b> crisps		<b>American</b> potato chips <i>/pə'teɪtəʊ 'tʃɪps/</i>
<b>British</b> biscuit		<b>American</b> cookie <i>/'kʊki/</i>
<b>British</b> peckish		<b>American</b> hungry <i>/'hʌŋgri/</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](http://americanenglish.state.gov)

<u>British</u>		<u>American</u>
kerb		curb
plough		plow
mould		mold
liquorice		licorice
cosy		cozy

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

ABCDEF GHIJKLMNOPQRSTUVWXYZ

# PTC Tasting Experiment

# Genetics of PTC Tasting

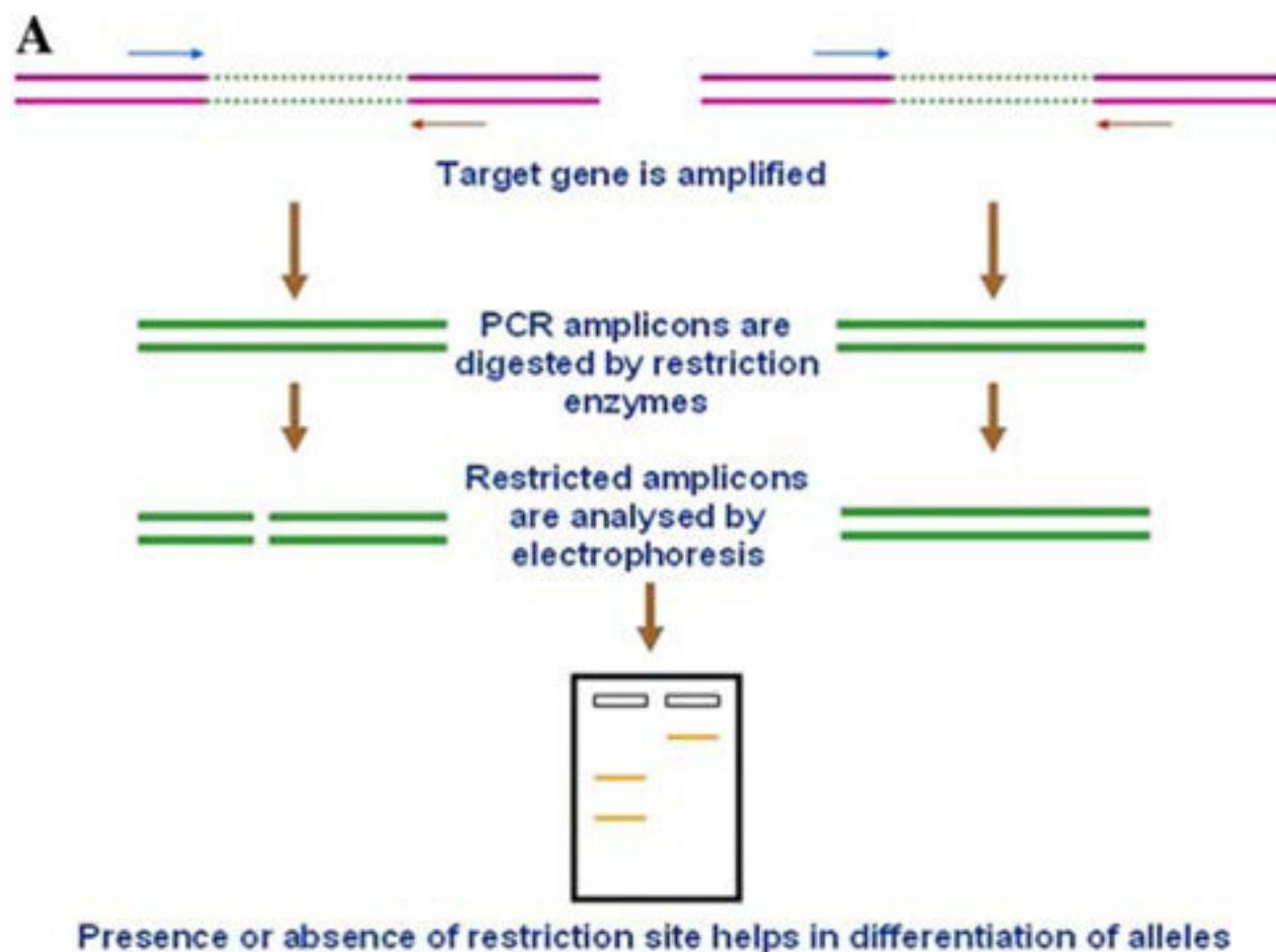
- PTC = phenylthiocarbamide
- Prepared by Arthur Fox at Du Pont Company in late 1920s
- Lab partner C.R. Noller complained of bitter taste
- Fox had no taste
- Followed up by Albert Blakeslee at Carnegie Department of Genetics
- Published in 1932



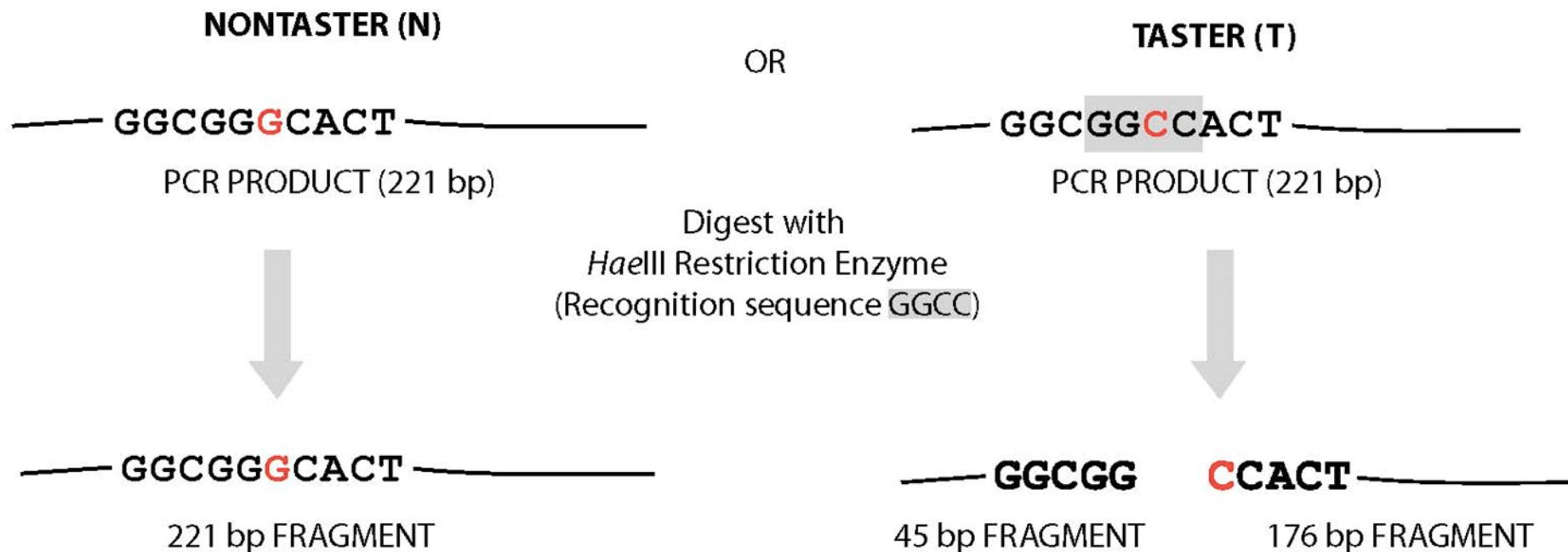
# Molecular Genetics of PTC Tasting

- 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

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