

Human Polymorphisms

PTC Tasting

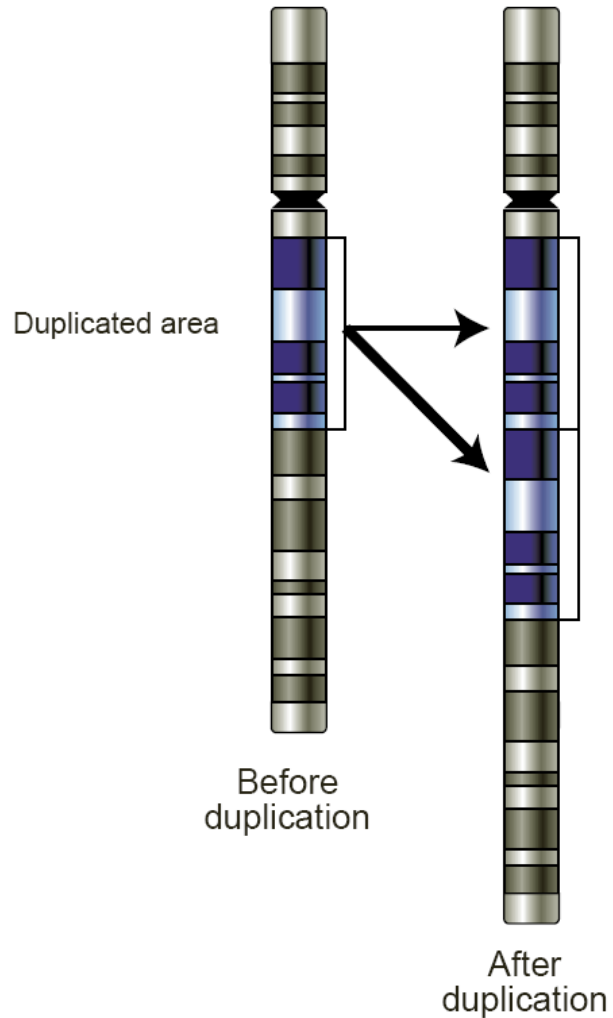
Variation



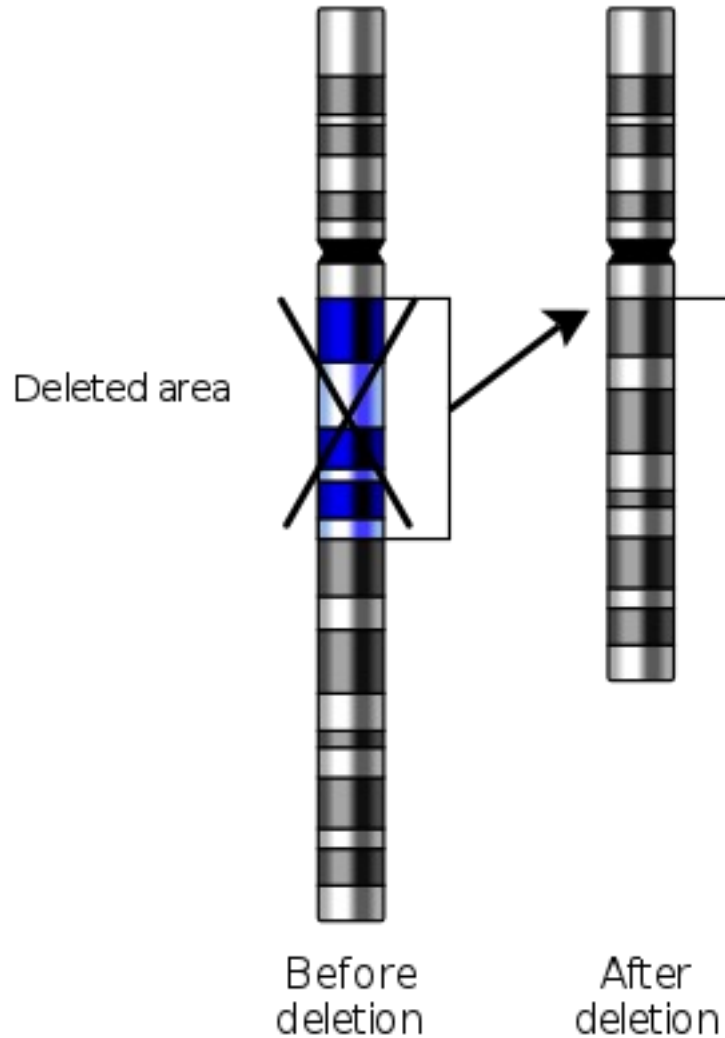
Genetic Variations

Copy Number Variation

- Different number of copies of the same gene



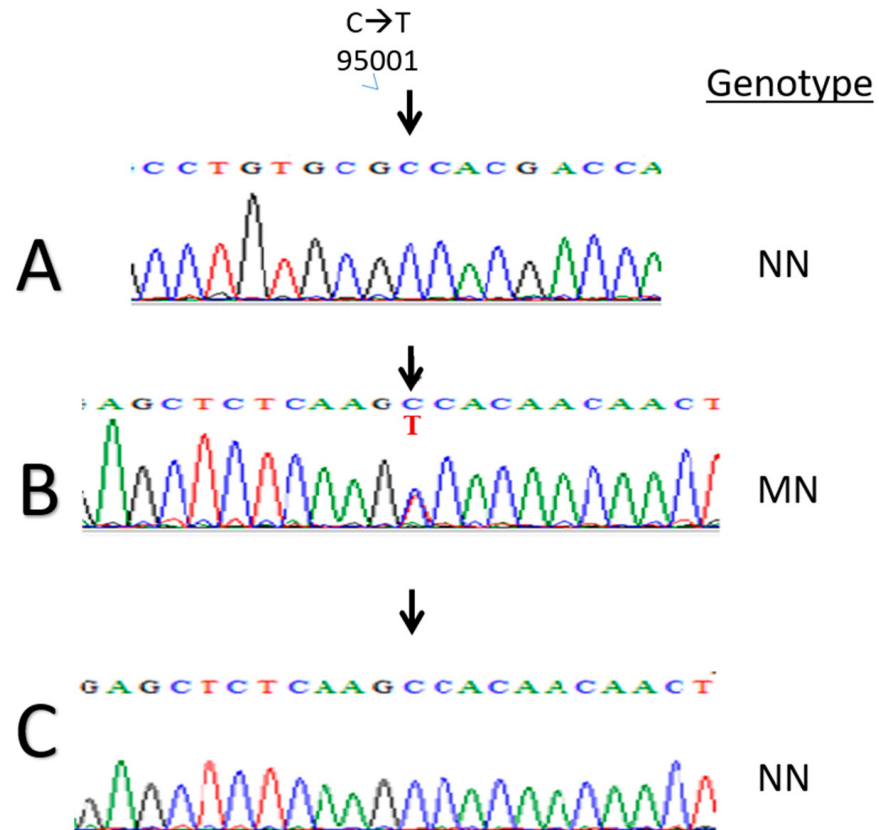
Genetic Variations



Copy Number Variation

- Genes may even get deleted

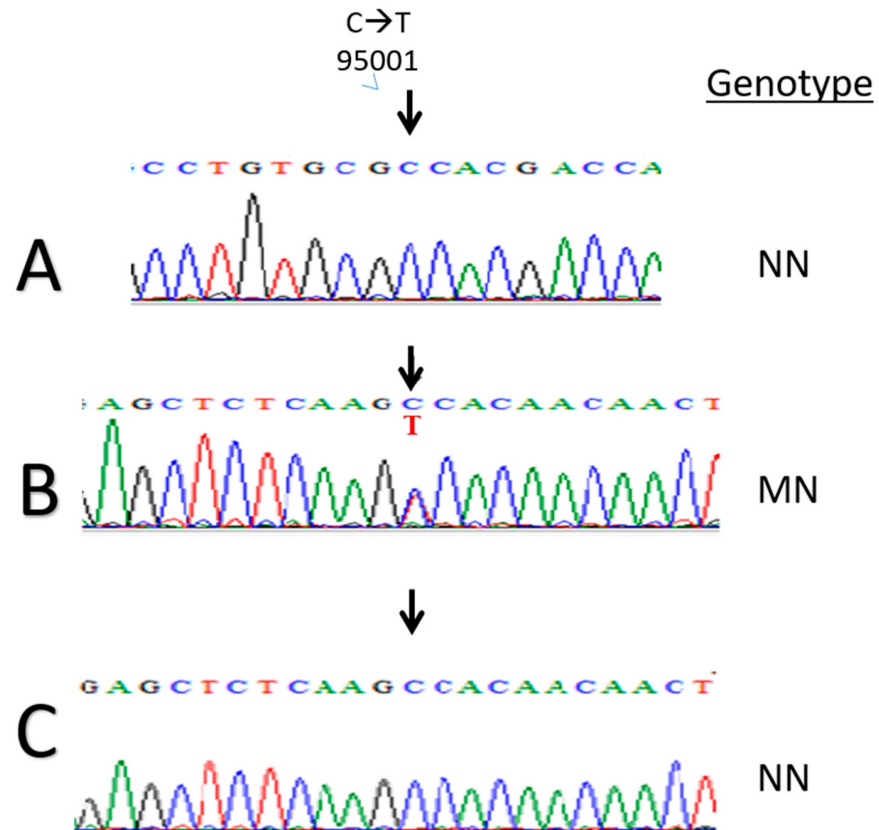
Genetic Variations



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



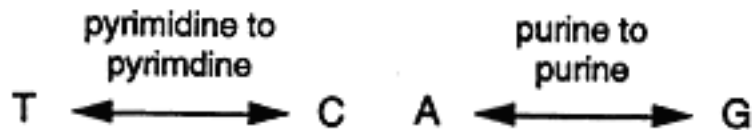
Polymorphism

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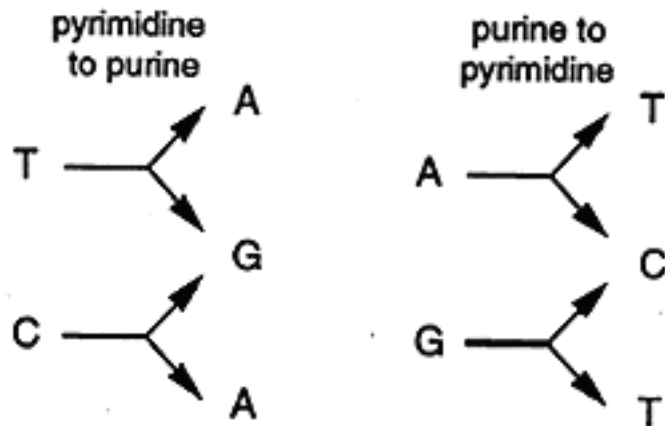
SNP Classifications

Transitions and Transversions

Transitions:



Transversions:

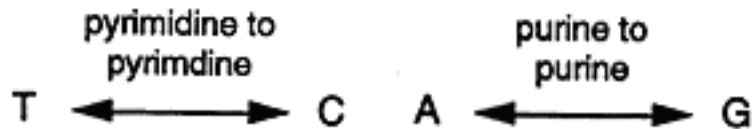


- Transition (more common)
 - a nucleotide is changed to another nucleotide with a similar chemical structure
- Transversion (less common)
 - a nucleotide is changed to another nucleotide that does not have the same chemical structure

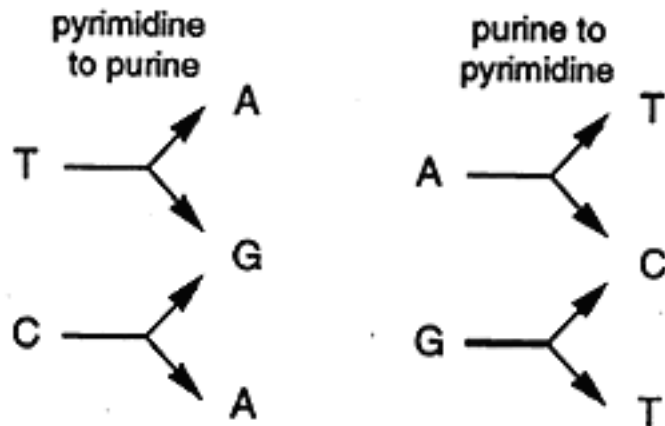
SNP Classifications

Transitions and Transversions

Transitions:



Transversions:








- Cytosine, Thymine –
(pyrimidines)

- Adenine, Guanine –
(purine)

Most variations are subtle (phonological)



British & American English 		
<small>americanenglish.state.gov</small>		
British chips		American french fries <small>/ˈfrɛntʃ ˈfraɪz/</small>
British crisps		American potato chips <small>/pəˈteɪtəʊ ˈtʃɪps/</small>
British biscuit		American cookie <small>/ˈkʊki/</small>
British peckish		American hungry <small>/ˈhʌŋɡri/</small>

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A E American English at State 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

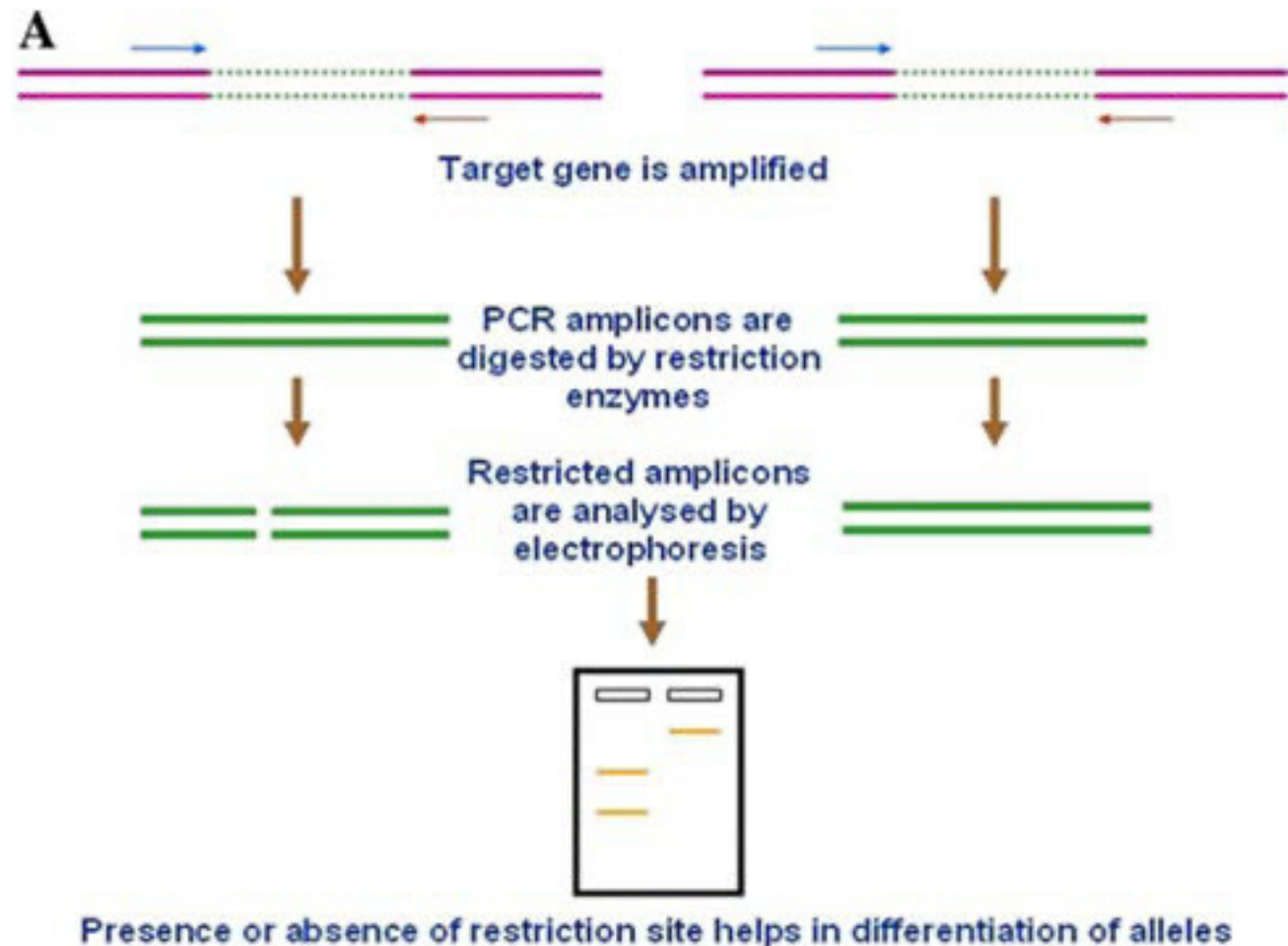
- PTC = phenylthiocarbamide
- Prepared by Arthur Fox at Du Pont Company in late 1920s
- Lab partner C.R. Noller complained of bitter taste
- Fox had no taste
- Followed up by Albert Blakeslee at Carnegie Department of Genetics
- Published in 1932



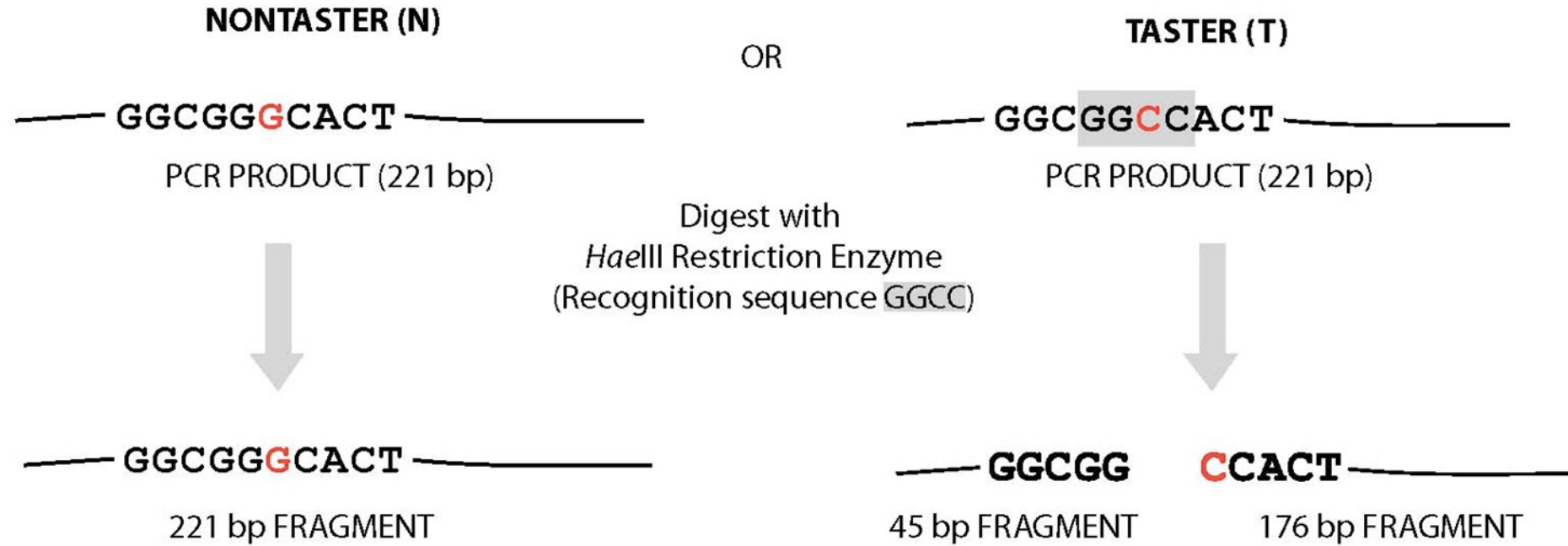
Molecular Genetics of PTC Tasting

- 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)