

GO:0033038 : bitter taste receptor activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0033038>)

GO - Biological Process

GO:0007186 : G protein-coupled receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007186>)

GO:0001580 : detection of chemical stimulus involved in sensory perception of bitter taste
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001580>)

GO - Cellular Component

GO:0016021 : integral component of membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0016021>)

GO:0005886 : plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005886>)

Presumptive Null

No ([#gpepbase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive Null=))

Molecular Type

Coding ([#gpepbase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular Type=))

Aberration Type

SNP ([#gpepbase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration Type=))

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

P49A and/or A262V and/or V296I

Experimental Evidence

Candidate Gene ([#gpepbase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental Evidence=))

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

Main Reference

Positional cloning of the human quantitative trait locus underlying taste sensitivity to phenylthiocarbamide. (2003) (<https://pubmed.ncbi.nlm.nih.gov/12595690>)

Authors

Kim UK; Jorgenson E; Coon H; Leppert M; Risch N; Drayna D

Abstract

The ability to taste the substance phenylthiocarbamide (PTC) has been widely used for genetic and anthropological studies, but genetic studies have produced conflicting results and demonstrated complex inheritance for this trait. We have identified a small region on chromosome 7q that shows strong linkage disequilibrium between single-nucleotide polymorphism (SNP) markers and PTC taste sensitivity in unrelated subjects. This region contains a single gene that encodes a member of the TAS2R bitter taste receptor family. We identified three coding SNPs giving rise to five haplotypes in this gene worldwide. These haplotypes completely explain the bimodal distribution of PTC taste sensitivity, thus accounting for the inheritance of the classically defined taste insensitivity and for 55 to 85% of the variance in PTC sensitivity. Distinct phenotypes were associated with specific haplotypes, which demonstrates that this gene has a direct influence on PTC taste sensitivity and that sequence variants at different sites interact with each other within the encoded gene product.

Additional References

Independent evolution of bitter-taste sensitivity in humans and chimpanzees. (2006) (<https://pubmed.ncbi.nlm.nih.gov/16612383>)

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS