GEPHE SUMMARY

Gephebase Gene

OR7D4 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^OR7D4^#gephebase-summary-title)

GepheID

GP00000800

Entry Status

Published

Main curator

Martin

PHENOTYPIC CHANGE

Trait Category

Physiology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Physiology^#gephebase-summary-title)

Trait

Olfaction (https://www.gephebase.org/search-criteria?/and+Trait=^Olfaction^#gephebase-summary-title)

Trait State in Taxon A

Homo sapiens sensitive to androstenone and androstadienone

Trait State in Taxon B

Homo sapiens less sensitive to androstenone and androstadienone

Ancestral State

Data not curated

Taxonomic Status

Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Intraspecific^#gephebase-summary-title)

Taxon A

Latin Name

Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Homo sapiens^#gephebase-summary-title)

Common Name

human

Synonyms

human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapian; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens

Rank

species

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Primates; Haplorrhini; Simiiformes; Catarrhini; Hominoidea; Hominidae; Homininae; Homo

Parent

Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)

NCBI Taxonomy ID

9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Homo sapiens^#gephebase-summary-title)

Common Name

human

Synonyms

human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapian; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens

Rank

species

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Primates; Haplorrhini; Simiiformes; Catarrhini; Hominoidea; Hominidae; Homininae; Homo

Parent

Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)

NCBI Taxonomy ID

9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)

is Taxon B an Infraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

OR7D4

Synonyms

OR19B; hg105; OR19-7; OR19-B; OR7D4P

String

9606.ENSP00000310488 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000310488)

Sequence Similarities

Belongs to the G-protein coupled receptor 1 family.

GO - Molecular Function

GO:0004930 : G protein-coupled receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004930)
GO:0004984 : olfactory receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004984)

UniProtKB Homo sapiens

Q8NG98 (http://www.uniprot.org/uniprot/Q8NG98)

GenebankID or UniProtKB

BC137147 (https://www.ncbi.nlm.nih.gov/nuccore/BC137147)

GO - Biological Process

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

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

Molecular Type

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

Aberration Type

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

R227G

Experimental Evidence

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

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

Main Reference

Genetic variation in a human odorant receptor alters odour perception. (2007) (https://pubmed.ncbi.nlm.nih.gov/17873857)

Authors

Keller A; Zhuang H; Chi Q; Vosshall LB; Matsunami H

Abstract

Human olfactory perception differs enormously between individuals, with large reported perceptual variations in the intensity and pleasantness of a given odour. For instance, androstenone (5alpha-androst-16-en-3-one), an odorous steroid derived from testosterone, is variously perceived by different individuals as offensive ("sweaty, urinous"), pleasant ("sweet, floral") or odourless. Similar variation in odour perception has been observed for several other odours. The mechanistic basis of variation in odour perception between individuals is unknown. We investigated whether genetic variation in human odorant receptor genes accounts in part for variation in odour perception between individuals. Here we show that a human odorant receptor, OR7D4, is selectively activated in vitro by androstenone and the related odorous steroid androstadienone (androsta-4,16-dien-3-one) and does not respond to a panel of 64 other odours and two solvents. A common variant of this receptor (OR7D4 WM) contains two non-synonymous single nucleotide polymorphisms (SNPs), resulting in two amino acid substitutions (R88W, T133M; hence 'RT') that severely impair function in vitro. Human subjects with RT/WM or WM/WM genotypes as a group were less sensitive to androstenone and androstadienone and found both odours less unpleasant than the RT/RT group. Genotypic variation in OR7D4 accounts for a significant proportion of the valence (pleasantness or unpleasantness) and intensity variance in perception of these steroidal odours. Our results demonstrate the first link between the function of a human odorant receptor in vitro and odour perception.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

5 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^OR7D4^/and+Taxon ID=^9606^/or+Gene Gephebase=^OR7D4^/and+Taxon ID=^9606^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS