

GEPHE SUMMARY

| | | | |
|---|----------------|------------|--------------|
| OR7D4 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^OR7D4^#gephebase-summary-title) | Gephebase Gene | GP00000801 | GepheID |
| Published | Entry Status | Martin | Main curator |

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 | Taxon B | Latin Name |
|--|-----------------------------|--|-----------------------------|
| Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Homo+sapiens^#gephebase-summary-title) | Common Name | Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Homo+sapiens^#gephebase-summary-title) | Common Name |
| human | Synonyms | human | Synonyms |
| human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapien; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Homo sapiens | | human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapien; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Homo sapiens | |
| species | Rank | species | Rank |
| 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 | 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 | Lineage |
| Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605) | Parent | Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605) | Parent |
| 9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606) | NCBI Taxonomy ID | 9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606) | NCBI Taxonomy ID |
| No | is Taxon A an Infraspecies? | No | is Taxon B an Infraspecies? |

GENOTYPIC CHANGE

| | | |
|--|-------------------------|--|
| OR7D4 | Generic Gene Name | UniProtKB Homo sapiens |
| OR19B; hg105; OR19-7; OR19-B; OR7D4P | Synonyms | GenebankID or UniProtKB |
| 9606.ENSP00000310488 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000310488) | String | BC137147 (https://www.ncbi.nlm.nih.gov/nuccore/BC137147) |
| Belongs to the G-protein coupled receptor 1 family. | Sequence Similarities | |
| 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) | GO - Molecular Function | |

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

T133M

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 (α -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