

GEPHE SUMMARY

|                                                                                                                                                                                                                                                                                                                     |                |              |
|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------|--------------|
|                                                                                                                                                                                                                                                                                                                     | Gephebase Gene | GepheID      |
| catechol-O-methyltransferase (COMT) ( <a +catechol-o-methyltransferase+(comt)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene Gephebase="catechol-O-methyltransferase (COMT)"#gephebase-summary-title</a> ) | GP00000171     |              |
|                                                                                                                                                                                                                                                                                                                     | Martin         | Main curator |
| Published                                                                                                                                                                                                                                                                                                           | Entry Status   |              |

PHENOTYPIC CHANGE

|                                                                                                                                                                                                                                                                                                                                                                                  |                             |                                                                                                                                                                                                                                                                                                                                                                                  |                             |
|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-----------------------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-----------------------------|
|                                                                                                                                                                                                                                                                                                                                                                                  | Trait Category              |                                                                                                                                                                                                                                                                                                                                                                                  |                             |
| Behavior ( <a +behavior+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait Category="Behavior"#gephebase-summary-title</a> )                                                                                                                                               |                             | Trait                                                                                                                                                                                                                                                                                                                                                                            |                             |
| Pain sensitivity ( <a +pain+sensitivity+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="Pain sensitivity"#gephebase-summary-title</a> )                                                                                                                                         |                             |                                                                                                                                                                                                                                                                                                                                                                                  |                             |
|                                                                                                                                                                                                                                                                                                                                                                                  | Trait State in Taxon A      |                                                                                                                                                                                                                                                                                                                                                                                  |                             |
| Homo sapiens                                                                                                                                                                                                                                                                                                                                                                     |                             | Trait State in Taxon B                                                                                                                                                                                                                                                                                                                                                           |                             |
| Homo sapiens high pain sensitive (HPS)                                                                                                                                                                                                                                                                                                                                           |                             |                                                                                                                                                                                                                                                                                                                                                                                  |                             |
|                                                                                                                                                                                                                                                                                                                                                                                  | Ancestral State             |                                                                                                                                                                                                                                                                                                                                                                                  |                             |
| Data not curated                                                                                                                                                                                                                                                                                                                                                                 |                             | Taxonomic Status                                                                                                                                                                                                                                                                                                                                                                 |                             |
| Intraspecific ( <a +intraspecific+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic Status="Intraspecific"#gephebase-summary-title</a> )                                                                                                                            |                             |                                                                                                                                                                                                                                                                                                                                                                                  |                             |
|                                                                                                                                                                                                                                                                                                                                                                                  | Taxon A                     |                                                                                                                                                                                                                                                                                                                                                                                  | Taxon B                     |
|                                                                                                                                                                                                                                                                                                                                                                                  | Latin Name                  |                                                                                                                                                                                                                                                                                                                                                                                  | Latin Name                  |
| Homo sapiens ( <a +homo+sapiens+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms="Homo sapiens"#gephebase-summary-title</a> )                                                                                                                           |                             | Homo sapiens ( <a +homo+sapiens+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms="Homo sapiens"#gephebase-summary-title</a> )                                                                                                                           |                             |
|                                                                                                                                                                                                                                                                                                                                                                                  | Common Name                 |                                                                                                                                                                                                                                                                                                                                                                                  | Common Name                 |
| human                                                                                                                                                                                                                                                                                                                                                                            |                             | human                                                                                                                                                                                                                                                                                                                                                                            |                             |
|                                                                                                                                                                                                                                                                                                                                                                                  | Synonyms                    |                                                                                                                                                                                                                                                                                                                                                                                  | 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                                                                                                                                                   |                             | 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                        |                                                                                                                                                                                                                                                                                                                                                                                  | Rank                        |
| species                                                                                                                                                                                                                                                                                                                                                                          |                             | species                                                                                                                                                                                                                                                                                                                                                                          |                             |
|                                                                                                                                                                                                                                                                                                                                                                                  | Lineage                     |                                                                                                                                                                                                                                                                                                                                                                                  | 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 |                             | 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                      |                                                                                                                                                                                                                                                                                                                                                                                  | Parent                      |
| Homo () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605</a> )                                                                                                                                                                                                      |                             | Homo () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605</a> )                                                                                                                                                                                                      |                             |
|                                                                                                                                                                                                                                                                                                                                                                                  | NCBI Taxonomy ID            |                                                                                                                                                                                                                                                                                                                                                                                  | NCBI Taxonomy ID            |
| 9606 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606</a> )                                                                                                                                                                                                                         |                             | 9606 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606</a> )                                                                                                                                                                                                                         |                             |
|                                                                                                                                                                                                                                                                                                                                                                                  | is Taxon A an Intraspecies? |                                                                                                                                                                                                                                                                                                                                                                                  | is Taxon B an Intraspecies? |
| No                                                                                                                                                                                                                                                                                                                                                                               |                             | No                                                                                                                                                                                                                                                                                                                                                                               |                             |

GENOTYPIC CHANGE

|                                                                                                                                                                                                                              |                         |                                                                                                                        |
|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|-------------------------|------------------------------------------------------------------------------------------------------------------------|
|                                                                                                                                                                                                                              | Generic Gene Name       | UniProtKB Homo sapiens                                                                                                 |
| COMT                                                                                                                                                                                                                         |                         | P21964 ( <a href="http://www.uniprot.org/uniprot/P21964">http://www.uniprot.org/uniprot/P21964</a> )                   |
|                                                                                                                                                                                                                              | Synonyms                | GenebankID or UniProtKB                                                                                                |
| HEL-S-98n                                                                                                                                                                                                                    |                         | AC005663 ( <a href="https://www.ncbi.nlm.nih.gov/nuccore/AC005663">https://www.ncbi.nlm.nih.gov/nuccore/AC005663</a> ) |
|                                                                                                                                                                                                                              | String                  |                                                                                                                        |
| 9606.ENSPO0000354511 ( <a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000354511">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000354511</a> ) |                         |                                                                                                                        |
|                                                                                                                                                                                                                              | Sequence Similarities   |                                                                                                                        |
| Belongs to the class I-like SAM-binding methyltransferase superfamily. Cation-dependent O-methyltransferase family.                                                                                                          |                         |                                                                                                                        |
|                                                                                                                                                                                                                              | GO - Molecular Function |                                                                                                                        |
| GO:0016206 : catechol O-methyltransferase activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0016206">https://www.ebi.ac.uk/QuickGO/term/GO:0016206</a> )                                                             |                         |                                                                                                                        |

GO:0102084 : L-dopa O-methyltransferase activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0102084>)  
GO:0000287 : magnesium ion binding  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000287>)  
GO:0008168 : methyltransferase activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008168>)  
GO:0008171 : O-methyltransferase activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008171>)  
GO:0102938 : orcinol O-methyltransferase activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0102938>)

GO - Biological Process

GO:0042424 : catecholamine catabolic process  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042424>)  
GO:0016036 : cellular response to phosphate starvation  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0016036>)  
GO:0032502 : developmental process  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032502>)  
GO:0042420 : dopamine catabolic process  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042420>)  
GO:0042417 : dopamine metabolic process  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042417>)  
GO:0008210 : estrogen metabolic process  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008210>)  
GO:0007565 : female pregnancy (<https://www.ebi.ac.uk/QuickGO/term/GO:0007565>)  
GO:0007612 : learning (<https://www.ebi.ac.uk/QuickGO/term/GO:0007612>)  
GO:0032259 : methylation (<https://www.ebi.ac.uk/QuickGO/term/GO:0032259>)  
GO:0048609 : multicellular organismal reproductive process  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048609>)  
GO:0045963 : negative regulation of dopamine metabolic process  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045963>)  
GO:0035814 : negative regulation of renal sodium excretion  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035814>)  
GO:0048662 : negative regulation of smooth muscle cell proliferation  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048662>)  
GO:0042135 : neurotransmitter catabolic process  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042135>)  
GO:0050668 : positive regulation of homocysteine metabolic process  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0050668>)  
GO:0051930 : regulation of sensory perception of pain  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051930>)  
GO:0042493 : response to drug (<https://www.ebi.ac.uk/QuickGO/term/GO:0042493>)  
GO:0043627 : response to estrogen (<https://www.ebi.ac.uk/QuickGO/term/GO:0043627>)  
GO:0032496 : response to lipopolysaccharide  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032496>)  
GO:0014070 : response to organic cyclic compound  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0014070>)  
GO:0048265 : response to pain (<https://www.ebi.ac.uk/QuickGO/term/GO:0048265>)  
GO:0007614 : short-term memory (<https://www.ebi.ac.uk/QuickGO/term/GO:0007614>)

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>)  
GO:0005829 : cytosol (<https://www.ebi.ac.uk/QuickGO/term/GO:0005829>)  
GO:0016020 : membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0016020>)  
GO:0070062 : extracellular exosome (<https://www.ebi.ac.uk/QuickGO/term/GO:0070062>)  
GO:0030425 : dendrite (<https://www.ebi.ac.uk/QuickGO/term/GO:0030425>)  
GO:0030424 : axon (<https://www.ebi.ac.uk/QuickGO/term/GO:0030424>)  
GO:0044297 : cell body (<https://www.ebi.ac.uk/QuickGO/term/GO:0044297>)  
GO:0043197 : dendritic spine (<https://www.ebi.ac.uk/QuickGO/term/GO:0043197>)  
GO:0043231 : intracellular membrane-bounded organelle  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043231>)  
GO:0045211 : postsynaptic membrane  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045211>)

Presumptive Null

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

Molecular Type

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

Aberration Type

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

Molecular Details of the Mutation

G/C - The synonymous mutation(s) affect mRNA stability and loop structures

Experimental Evidence

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

Main Reference

Human catechol-O-methyltransferase haplotypes modulate protein expression by altering mRNA secondary structure. (2006) (<https://pubmed.ncbi.nlm.nih.gov/17185601>)

Authors

Nackley AG; Shabalina SA; Tchivileva IE; Satterfield K; Korchynskiy O; Makarov SS; Maixner W; Diatchenko L

Abstract

Catechol-O-methyltransferase (COMT) is a key regulator of pain perception, cognitive function, and affective mood. Three common haplotypes of the human COMT gene, divergent in two synonymous and one nonsynonymous position, code for differences in COMT enzymatic activity and are associated with pain sensitivity. Haplotypes divergent in synonymous changes exhibited the largest difference in COMT enzymatic activity, due to a reduced amount of translated protein. The major COMT haplotypes varied with respect to messenger RNA local stem-loop structures, such that the most stable structure was associated with the lowest protein levels and enzymatic activity. Site-directed mutagenesis that eliminated the stable structure restored the amount of translated protein. These data highlight the functional significance of synonymous variations and suggest the importance of haplotypes over single-nucleotide polymorphisms for analysis of genetic variations.

Additional References

RELATED GEPHE

No matches found.

Related Genes

No matches found.

Related Haplotypes

EXTERNAL LINKS

COMMENTS