

GEPHE SUMMARY

<p>GADD45G (https://www.gephebase.org/search-criteria?/and+GeneGephebase=GADD45G#gephebase-summary-title)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00000384</p> <p>Martin</p>	<p>GepheID</p> <p>Main curator</p>
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PHENOTYPIC CHANGE

<p>Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=Morphology#gephebase-summary-title)</p> <p>Brain development (https://www.gephebase.org/search-criteria?/and+Trait=Braindevelopment#gephebase-summary-title)</p> <p>Homo sapiens</p> <p>Pan troglodytes</p> <p>Taxon A</p> <p>Interspecific (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=Interspecific#gephebase-summary-title)</p>	<p>Trait Category</p> <p>Trait</p> <p>Trait State in Taxon A</p> <p>Trait State in Taxon B</p> <p>Ancestral State</p> <p>Taxonomic Status</p>	<p>Taxon A</p> <p>Latin Name</p> <p>Homo sapiens (https://www.gephebase.org/search-criteria?/and+TaxonandSynonyms=HomoSapiens#gephebase-summary-title)</p> <p>Common Name</p> <p>human</p> <p>Synonyms</p> <p>human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sapiens; Homo sapeins; Homo sapien; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens</p> <p>Rank</p> <p>species</p> <p>Lineage</p> <p>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</p> <p>Parent</p> <p>Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)</p> <p>NCBI Taxonomy ID</p> <p>9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)</p> <p>No</p> <p>is Taxon A an Infrappecies?</p>	<p>Taxon B</p> <p>Latin Name</p> <p>Pan troglodytes (https://www.gephebase.org/search-criteria?/and+TaxonandSynonyms=PanTroglodytes#gephebase-summary-title)</p> <p>Common Name</p> <p>chimpanzee</p> <p>Synonyms</p> <p>chimpanzee; Chimpansee troglodytes</p> <p>Rank</p> <p>species</p> <p>Lineage</p> <p>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; Pan</p> <p>Parent</p> <p>Pan (chimpanzees) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9596)</p> <p>NCBI Taxonomy ID</p> <p>9598 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9598)</p> <p>No</p> <p>is Taxon B an Infrappecies?</p>
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GENOTYPIC CHANGE

<p>GADD45G</p> <p>CR6; DDIT2; GRP17; GADD45gamma</p> <p>9606.ENSPO0000252506 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSPO0000252506)</p> <p>Belongs to the GADD45 family.</p> <p>-</p> <p>GO:0007275 : multicellular organism development (https://www.ebi.ac.uk/QuickGO/term/GO:0007275)</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p> <p>GO - Biological Process</p>	<p>UniProtKB Homo sapiens</p> <p>O95257 (http://www.uniprot.org/uniprot/O95257)</p> <p>GenebankID or UniProtKB</p> <p>JAA34093 (https://www.ncbi.nlm.nih.gov/nucleotide/JAA34093)</p>
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GO:0030154 : cell differentiation (<https://www.ebi.ac.uk/QuickGO/term/GO:0030154>)
 GO:0006915 : apoptotic process (<https://www.ebi.ac.uk/QuickGO/term/GO:0006915>)
 GO:0046330 : positive regulation of JNK cascade
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0046330>)
 GO:0120162 : positive regulation of cold-induced thermogenesis
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0120162>)
 GO:0043065 : positive regulation of apoptotic process
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0043065>)
 GO:0000185 : activation of MAPKKK activity
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0000185>)
 GO:1900745 : positive regulation of p38MAPK cascade
 (<https://www.ebi.ac.uk/QuickGO/term/GO:1900745>)
 GO:0051726 : regulation of cell cycle (<https://www.ebi.ac.uk/QuickGO/term/GO:0051726>)

GO - Cellular Component

GO:0005737 : cytoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005737>)
 GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)

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+No^#gephebase-summary-title))

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=Cis-regulatory^#gephebase-summary-title))

Aberration Type

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

Deletion Size

10-100 kb

Molecular Details of the Mutation

Enhancer loss

Experimental Evidence

Association Mapping ([https://www.gephebase.org/search-criteria?/and+Experimental Evidence=Association Mapping^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=Association+Mapping^#gephebase-summary-title))

Main Reference

Human-specific loss of regulatory DNA and the evolution of human-specific traits. (2011) (<https://pubmed.ncbi.nlm.nih.gov/21390129>)

Authors

McLean CY; Reno PL; Pollen AA; Bassan AI; Capellini TD; Guenther C; Indjeian VB; Lim X; Menke DB; Schaar BT; Wenger AM; Bejerano G; Kingsley DM

Abstract

Humans differ from other animals in many aspects of anatomy, physiology, and behaviour; however, the genotypic basis of most human-specific traits remains unknown. Recent whole-genome comparisons have made it possible to identify genes with elevated rates of amino acid change or divergent expression in humans, and non-coding sequences with accelerated base pair changes. Regulatory alterations may be particularly likely to produce phenotypic effects while preserving viability, and are known to underlie interesting evolutionary differences in other species. Here we identify molecular events particularly likely to produce significant regulatory changes in humans: complete deletion of sequences otherwise highly conserved between chimpanzees and other mammals. We confirm 510 such deletions in humans, which fall almost exclusively in non-coding regions and are enriched near genes involved in steroid hormone signalling and neural function. One deletion removes a sensory vibrissae and penile spine enhancer from the human androgen receptor (AR) gene, a molecular change correlated with anatomical loss of androgen-dependent sensory vibrissae and penile spines in the human lineage. Another deletion removes a forebrain subventricular zone enhancer near the tumour suppressor gene growth arrest and DNA-damage-inducible, gamma (GADD45G), a loss correlated with expansion of specific brain regions in humans. Deletions of tissue-specific enhancers may thus accompany both loss and gain traits in the human lineage, and provide specific examples of the kinds of regulatory alterations and inactivation events long proposed to have an important role in human evolutionary divergence.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS