GEPHE SUMMARY

Gephebase Gene
GADD45G (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^GADD45G^#gephebase-summary-title)

GepheID
GP00000384

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

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

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

Trait State in Taxon A
Homo sapiens

Trait State in Taxon B
Pan troglodytes

Ancestral State
Taxon A

Taxonomic Status
Interspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Interspecific^#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
Pan troglodytes (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Pan troglodytes^#gephebase-summary-title)

Common Name
chimpanzee

Synonyms
chimpanzee; Chimpansee troglodytes

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; Pan

Parent
Pan (chimpanzees) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9596)

NCBI Taxonomy ID
9598 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9598)

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
GADD45G

Synonyms
CR6; DDIT2; GRP17; GADD45gamma

String
9606.ENSP00000252506 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000252506)

Sequence Similarities
Belongs to the GADD45 family.

GO - Molecular Function
-

GO - Biological Process
GO:0007275 : multicellular organism development (https://www.ebi.ac.uk/QuickGO/term/GO:0007275)

UniProtKB Homo sapiens
O95257 (http://www.uniprot.org/uniprot/O95257)

GenebankID or UniProtKB
JAA34093 (https://www.ncbi.nlm.nih.gov/nuccore/JAA34093)

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)

Molecular Type

Cis-regulatory (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)

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)

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