

GEPHE SUMMARY

Gephebase Gene		GepheID
AR (<a +ar+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+AR+"#gephebase-summary-title)	GP00000102	
Entry Status		Main curator
Published		Martin

PHENOTYPIC CHANGE

Trait #1	Trait Category
Morphology (<a +morphology+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Morphology+"#gephebase-summary-title)	
Sensory vibrissae (loss) (<a +sensory+vibrissae+(loss)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Sensory+vibrissae+(loss)+"#gephebase-summary-title)	
Pan troglodytes	
Homo sapiens	

Trait #2	Trait Category
Morphology (<a +morphology+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Morphology+"#gephebase-summary-title)	
Penile spine (loss) (<a +penile+spine+(loss)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Penile+spine+(loss)+"#gephebase-summary-title)	
-	
-	

Ancestral State	
Taxon A	
Taxonomic Status	
Interspecific (<a +interspecific+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status="+Interspecific+"#gephebase-summary-title)	

Taxon A		Taxon B	
Pan troglodytes		Homo sapiens	
(<a +pan+troglodytes+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Pan+troglodytes+"#gephebase-summary-title)		(<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)	
chimpanzee		human	
chimpanzee; Chimpansee troglodytes		human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapien; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens	
species		species	
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		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	
Pan (chimpanzees) - (Rank: genus)		Homo () - (Rank: genus)	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9596)		(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)	
9598		9606	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9598)		(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)	
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
No		No	

GENOTYPIC CHANGE

GENOTYPIC CHANGE		Generic Gene Name	UniProtKB Homo sapiens
AR		P10275 (http://www.uniprot.org/uniprot/P10275)	
		Synonyms	GenebankID or UniProtKB
KD; AIS; AR8; TFM; DHTR; SBMA; HYSPI; NR3C4; SMAX1; HUMARA		BAB59056 (https://www.ncbi.nlm.nih.gov/nuccore/BAB59056)	
		String	
9606.ENSPP00000363822			
(http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000363822)			
		Sequence Similarities	
Belongs to the nuclear hormone receptor family. NR3 subfamily.			
		GO - Molecular Function	
GO:0046983 : protein dimerization activity			
(https://www.ebi.ac.uk/QuickGO/term/GO:0046983)			
GO:0003700 : DNA-binding transcription factor activity			
(https://www.ebi.ac.uk/QuickGO/term/GO:0003700)			
GO:0008134 : transcription factor binding			
(https://www.ebi.ac.uk/QuickGO/term/GO:0008134)			
GO:0044212 : transcription regulatory region DNA binding			
(https://www.ebi.ac.uk/QuickGO/term/GO:0044212)			
GO:0008270 : zinc ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0008270)			
GO:0003677 : DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003677)			
GO:0005497 : androgen binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005497)			
GO:0004882 : androgen receptor activity			
(https://www.ebi.ac.uk/QuickGO/term/GO:0004882)			
GO:0051117 : ATPase binding (https://www.ebi.ac.uk/QuickGO/term/GO:0051117)			
GO:0008013 : beta-catenin binding (https://www.ebi.ac.uk/QuickGO/term/GO:0008013)			
GO:0003682 : chromatin binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003682)			
GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific			
(https://www.ebi.ac.uk/QuickGO/term/GO:0000981)			
GO:0019899 : enzyme binding (https://www.ebi.ac.uk/QuickGO/term/GO:0019899)			
GO:0004879 : nuclear receptor activity			
(https://www.ebi.ac.uk/QuickGO/term/GO:0004879)			
GO:0070974 : POU domain binding (https://www.ebi.ac.uk/QuickGO/term/GO:0070974)			
GO:0001077 : proximal promoter DNA-binding transcription activator activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0001077)			
GO:0001091 : RNA polymerase II basal transcription factor binding			
(https://www.ebi.ac.uk/QuickGO/term/GO:0001091)			
GO:0000978 : RNA polymerase II proximal promoter sequence-specific DNA binding			
(https://www.ebi.ac.uk/QuickGO/term/GO:0000978)			
GO:0001085 : RNA polymerase II transcription factor binding			
(https://www.ebi.ac.uk/QuickGO/term/GO:0001085)			
GO:0005102 : signaling receptor binding			
(https://www.ebi.ac.uk/QuickGO/term/GO:0005102)			
GO:0005496 : steroid binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005496)			
		GO - Biological Process	
GO:0007165 : signal transduction (https://www.ebi.ac.uk/QuickGO/term/GO:0007165)			
GO:0045944 : positive regulation of transcription by RNA polymerase II			
(https://www.ebi.ac.uk/QuickGO/term/GO:0045944)			
GO:0000122 : negative regulation of transcription by RNA polymerase II			
(https://www.ebi.ac.uk/QuickGO/term/GO:0000122)			
GO:0045893 : positive regulation of transcription, DNA-templated			
(https://www.ebi.ac.uk/QuickGO/term/GO:0045893)			
GO:0008283 : cell proliferation (https://www.ebi.ac.uk/QuickGO/term/GO:0008283)			
GO:0008284 : positive regulation of cell proliferation			
(https://www.ebi.ac.uk/QuickGO/term/GO:0008284)			
GO:0060520 : activation of prostate induction by androgen receptor signaling pathway			
(https://www.ebi.ac.uk/QuickGO/term/GO:0060520)			
GO:0030521 : androgen receptor signaling pathway			
(https://www.ebi.ac.uk/QuickGO/term/GO:0030521)			
GO:0048645 : animal organ formation			
(https://www.ebi.ac.uk/QuickGO/term/GO:0048645)			
GO:0007267 : cell-cell signaling (https://www.ebi.ac.uk/QuickGO/term/GO:0007267)			
GO:0071383 : cellular response to steroid hormone stimulus			
(https://www.ebi.ac.uk/QuickGO/term/GO:0071383)			
GO:0071394 : cellular response to testosterone stimulus			
(https://www.ebi.ac.uk/QuickGO/term/GO:0071394)			
GO:0060742 : epithelial cell differentiation involved in prostate gland development			
(https://www.ebi.ac.uk/QuickGO/term/GO:0060742)			
GO:0003382 : epithelial cell morphogenesis			
(https://www.ebi.ac.uk/QuickGO/term/GO:0003382)			
GO:0001701 : in utero embryonic development			
(https://www.ebi.ac.uk/QuickGO/term/GO:0001701)			
GO:0030522 : intracellular receptor signaling pathway			
(https://www.ebi.ac.uk/QuickGO/term/GO:0030522)			
GO:0060599 : lateral sprouting involved in mammary gland duct morphogenesis			

(<https://www.ebi.ac.uk/QuickGO/term/GO:0060599>)
GO:0033327 : Leydig cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0033327>)
GO:0048808 : male genitalia morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048808>)
GO:0019102 : male somatic sex determination
(<https://www.ebi.ac.uk/QuickGO/term/GO:0019102>)
GO:0060749 : mammary gland alveolus development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060749>)
GO:0060571 : morphogenesis of an epithelial fold
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060571>)
GO:0035264 : multicellular organism growth
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035264>)
GO:0008285 : negative regulation of cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008285>)
GO:0050680 : negative regulation of epithelial cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0050680>)
GO:2001237 : negative regulation of extrinsic apoptotic signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:2001237>)
GO:0045720 : negative regulation of integrin biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045720>)
GO:0045597 : positive regulation of cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045597>)
GO:0060769 : positive regulation of epithelial cell proliferation involved in prostate gland development (<https://www.ebi.ac.uk/QuickGO/term/GO:0060769>)
GO:0010628 : positive regulation of gene expression
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010628>)
GO:0043568 : positive regulation of insulin-like growth factor receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043568>)
GO:0045726 : positive regulation of integrin biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045726>)
GO:0033148 : positive regulation of intracellular estrogen receptor signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0033148>)
GO:0043410 : positive regulation of MAPK cascade
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043410>)
GO:0051092 : positive regulation of NF-kappaB transcription factor activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051092>)
GO:0042327 : positive regulation of phosphorylation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042327>)
GO:0045945 : positive regulation of transcription by RNA polymerase III
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045945>)
GO:0030850 : prostate gland development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030850>)
GO:0060740 : prostate gland epithelium morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060740>)
GO:0060736 : prostate gland growth (<https://www.ebi.ac.uk/QuickGO/term/GO:0060736>)
GO:0051259 : protein complex oligomerization
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051259>)
GO:0016579 : protein deubiquitination
(<https://www.ebi.ac.uk/QuickGO/term/GO:0016579>)
GO:0050790 : regulation of catalytic activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0050790>)
GO:0048638 : regulation of developmental growth
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048638>)
GO:1903076 : regulation of protein localization to plasma membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:1903076>)
GO:0003073 : regulation of systemic arterial blood pressure
(<https://www.ebi.ac.uk/QuickGO/term/GO:0003073>)
GO:0072520 : seminiferous tubule development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0072520>)
GO:0007548 : sex differentiation (<https://www.ebi.ac.uk/QuickGO/term/GO:0007548>)
GO:0007338 : single fertilization (<https://www.ebi.ac.uk/QuickGO/term/GO:0007338>)
GO:0007283 : spermatogenesis (<https://www.ebi.ac.uk/QuickGO/term/GO:0007283>)
GO:0060748 : tertiary branching involved in mammary gland duct morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0060748>)
GO:0006367 : transcription initiation from RNA polymerase II promoter
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006367>)
GO:0006351 : transcription, DNA-templated
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006351>)

GO - Cellular Component

GO:0005886 : plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005886>)
GO:0005737 : cytoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005737>)
GO:0005829 : cytosol (<https://www.ebi.ac.uk/QuickGO/term/GO:0005829>)
GO:0005654 : nucleoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005654>)
GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)
GO:0032991 : protein-containing complex
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032991>)
GO:0000790 : nuclear chromatin (<https://www.ebi.ac.uk/QuickGO/term/GO:0000790>)

GO:0016607 : nuclear speck (https://www.ebi.ac.uk/QuickGO/term/GO:0016607)	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
Candidate Gene (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Candidate Gene^#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

No matches found.	Related Genes
No matches found.	Related Haplotypes

EXTERNAL LINKS

COMMENTS