

GEPHE SUMMARY

<p>FoxP2 (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase+FoxP2+Gephebase-summary-title)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00000353</p> <p>Martin</p>	<p>GepheID</p> <p>Main curator</p>
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PHENOTYPIC CHANGE

<p>Behavior (https://www.gephebase.org/search-criteria?/and+Trait+Category+Behavior+Gephebase-summary-title)</p>	<p>Trait Category</p>			
<p>Gene expression change (transcriptional targets) (https://www.gephebase.org/search-criteria?/and+Trait+Gene+expression+change+(transcriptional+targets)+Gephebase-summary-title)</p>	<p>Trait</p>			
<p>Pan troglodytes</p>	<p>Trait State in Taxon A</p>			
<p>Homo sapiens</p>	<p>Trait State in Taxon B</p>			
<p>Taxon A</p>	<p>Ancestral State</p>			
<p>Interspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status+Interspecific+Gephebase-summary-title)</p>	<p>Taxonomic Status</p>			
<p>Taxon A</p>	<p>Latin Name</p>	<p>Taxon B</p>	<p>Latin Name</p>	
<p>Pan troglodytes (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+Pan+troglodytes+Gephebase-summary-title)</p>	<p>Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms+Homo+sapiens+Gephebase-summary-title)</p>			
<p>chimpanzee</p>	<p>Common Name</p>	<p>human</p>	<p>Common Name</p>	
<p>chimpanzee; Chimpansee troglodytes</p>	<p>Synonyms</p>	<p>human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins;</p>	<p>Synonyms</p>	
<p>species</p>	<p>Rank</p>	<p>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>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>Lineage</p>	<p>species</p>	<p>Lineage</p>	
<p>Pan (chimpanzees) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9596)</p>	<p>Parent</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>9598 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9598)</p>	<p>NCBI Taxonomy ID</p>	<p>Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)</p>	<p>NCBI Taxonomy ID</p>	
<p>No</p>	<p>is Taxon A an Intraspecies?</p>	<p>9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)</p>	<p>is Taxon B an Intraspecies?</p>	
		<p>No</p>		

GENOTYPIC CHANGE

<p>FOXP2</p>	<p>Generic Gene Name</p>	<p>O15409 (http://www.uniprot.org/uniprot/O15409)</p>	<p>UniProtKB Homo sapiens</p>
<p>SPCH1; CAGH44; TNRC10</p>	<p>Synonyms</p>	<p>AF467255 (https://www.ncbi.nlm.nih.gov/nucleotide/AF467255)</p>	<p>GenebankID or UniProtKB</p>
<p>9606.ENSPO0000386200 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000386200)</p>	<p>String</p>		
<p>-</p>	<p>Sequence Similarities</p>		
<p>GO:0042802 : identical protein binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042802)</p> <p>GO:0042803 : protein homodimerization activity</p>	<p>GO - Molecular Function</p>		

(<https://www.ebi.ac.uk/QuickGO/term/GO:0042803>)
 GO:0003700 : DNA-binding transcription factor activity
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0003700>)
 GO:0046982 : protein heterodimerization activity
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0046982>)
 GO:0043565 : sequence-specific DNA binding
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0043565>)
 GO:0046872 : metal ion binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0046872>)
 GO:0003677 : DNA binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0003677>)
 GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0000981>)
 GO:0000978 : RNA polymerase II proximal promoter sequence-specific DNA binding
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0000978>)
 GO:0001078 : proximal promoter DNA-binding transcription repressor activity, RNA polymerase II-specific (<https://www.ebi.ac.uk/QuickGO/term/GO:0001078>)
 GO:0050681 : androgen receptor binding
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0050681>)

GO - Biological Process

GO:0045892 : negative regulation of transcription, DNA-templated
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0045892>)
 GO:0009791 : post-embryonic development
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0009791>)
 GO:0048286 : lung alveolus development
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0048286>)
 GO:0060501 : positive regulation of epithelial cell proliferation involved in lung morphogenesis (<https://www.ebi.ac.uk/QuickGO/term/GO:0060501>)
 GO:0002053 : positive regulation of mesenchymal cell proliferation
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0002053>)
 GO:0043010 : camera-type eye development
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0043010>)
 GO:0021757 : caudate nucleus development
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0021757>)
 GO:0021549 : cerebellum development
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0021549>)
 GO:0021987 : cerebral cortex development
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0021987>)
 GO:0098582 : innate vocalization behavior
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0098582>)
 GO:0021758 : putamen development (<https://www.ebi.ac.uk/QuickGO/term/GO:0021758>)
 GO:0033574 : response to testosterone
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0033574>)
 GO:0060013 : righting reflex (<https://www.ebi.ac.uk/QuickGO/term/GO:0060013>)
 GO:0007519 : skeletal muscle tissue development
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0007519>)
 GO:0048745 : smooth muscle tissue development
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0048745>)
 GO:0042297 : vocal learning (<https://www.ebi.ac.uk/QuickGO/term/GO:0042297>)

GO - Cellular Component

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

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

2 aa changes (T303N and N325S) resulting in changes of the regulated transcriptional targets in a human neuron cell line. Speculatively; these changes could be associated with language skills but these connections with null-mutant phenotypes are dubious - whether one or both mutations are required is unknown

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

Human-specific transcriptional regulation of CNS development genes by FOXP2. (2009) (<https://pubmed.ncbi.nlm.nih.gov/19907493>)

Authors

Konopka G; Bomar JM; Winden K; Coppola G; Jonsson ZO; Gao F; Peng S; Preuss TM; Wohlschlegel JA; Geschwind DH

Abstract

The signalling pathways controlling both the evolution and development of language in the human brain remain unknown. So far, the transcription factor FOXP2 (forkhead box P2) is the only gene implicated in Mendelian forms of human speech and language dysfunction. It has been proposed that the amino acid composition in the human variant of FOXP2 has undergone accelerated evolution, and this two-amino-acid change occurred around the time of language emergence in humans. However, this remains controversial, and whether the acquisition of these

amino acids in human FOXP2 has any functional consequence in human neurons remains untested. Here we demonstrate that these two human-specific amino acids alter FOXP2 function by conferring differential transcriptional regulation in vitro. We extend these observations in vivo to human and chimpanzee brain, and use network analysis to identify novel relationships among the differentially expressed genes. These data provide experimental support for the functional relevance of changes in FOXP2 that occur on the human lineage, highlighting specific pathways with direct consequences for human brain development and disease in the central nervous system (CNS). Because FOXP2 has an important role in speech and language in humans, the identified targets may have a critical function in the development and evolution of language circuitry in humans.

Additional References

Humanized Foxp2 accelerates learning by enhancing transitions from declarative to procedural performance. (2014) (<https://pubmed.ncbi.nlm.nih.gov/25225386>)

RELATED GEPHE

No matches found.

Related Genes

No matches found.

Related Haplotypes

EXTERNAL LINKS

COMMENTS