GEPHE SUMMARY

Gephebase Gene

ARHGAP11B (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^ARHGAP11B^#gephebase-summary-title)

Entry Status

Published

GepheID

GP00001442

Main curator

Prigent

PHENOTYPIC CHANGE

Trait Category

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

Trait

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

Trait State in Taxon A

ancestral human >500 000 years ago

Trait State in Taxon B

modern human and archaic hominins (Neanderthal and Denisova)

Ancestral State

Taxon A

Taxonomic Status

Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Intraspecific^#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

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 B an Infraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

ARHGAP11B

Synonyms

B'-T; FAM7B1; GAP (1-8)

String

9606.ENSP00000392760 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000392760)

Sequence Similarities

-

GO - Molecular Function

GO:0005096 : GTPase activator activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005096)

GO - Biological Process

UniProtKB Homo sapiens

Q3KRB8 (http://www.uniprot.org/uniprot/Q3KRB8)

GenebankID or UniProtKB

NM_001039841 (https://www.ncbi.nlm.nih.gov/nuccore/NM_001039841)

GO:0007165 : signal transduction (https://www.ebi.ac.uk/QuickGO/term/GO:0007165)
GO:0021987 : cerebral cortex development
(https://www.ebi.ac.uk/QuickGO/term/GO:0021987)
GO:0051056 : regulation of small GTPase mediated signal transduction
(https://www.ebi.ac.uk/QuickGO/term/GO:0051056)

GO - Cellular Component

GO:0005829 : cytosol (https://www.ebi.ac.uk/QuickGO/term/GO:0005829)

Presumptive Null

Yes (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Yes^#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

-

Molecular Details of the Mutation

c.661C>G new splice site mutation removing 55nt from exon 5 truncating a GAP domain

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

A single splice site mutation in human-specific ARHGAP11B causes basal progenitor amplification. (2016) (https://pubmed.ncbi.nlm.nih.gov/27957544)

Authors

Florio M; Namba T; PÃ¤Ã¤bo S; Hiller M; Huttner WB

Abstract

The gene ARHGAP11B promotes basal progenitor amplification and is implicated in neocortex expansion. It arose on the human evolutionary lineage by partial duplication of ARHGAP11A, which encodes a Rho guanosine triphosphatase-activating protein (RhoGAP). However, a lack of 55 nucleotides in ARHGAP11B mRNA leads to loss of RhoGAP activity by GAP domain truncation and addition of a human-specific carboxy-terminal amino acid sequence. We show that these 55 nucleotides are deleted by mRNA splicing due to a single Câ†'G substitution that creates a novel splice donor site. We reconstructed an ancestral ARHGAP11B complementary DNA without this substitution. Ancestral ARHGAP11B exhibits RhoGAP activity but has no ability to increase basal progenitors during neocortex development. Hence, a single nucleotide substitution underlies the specific properties of ARHGAP11B that likely contributed to the evolutionary expansion of the human neocortex.

Additional References

RELATED GEPHE

Related Genes

1 (FRIZZLED8) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9606^/and+Trait=Neocortex development/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@Splicing