GEPHE SUMMARY

Gephebase Gene
myosin heavy chain 16 (MYH16) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^myosin heavy chain 16 (MYH16)^#gephebase-summary-title)

GepheID
GP00000683

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

Trait State in Taxon A
Pan troglodytes

Trait State in Taxon B
Homo sapiens

Ancestral State
Taxon A

Taxonomic Status
Intergeneric or Higher (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Intergeneric or Higher^#gephebase-summary-title)

Taxon A

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 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
MYH16

Synonyms
MYH5

String
-

Sequence Similarities
Belongs to the TRAFAC class myosin-kinesin ATPase superfamily. Myosin family.

GO - Molecular Function
GO:0003774 : motor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0003774)

GO - Biological Process
-

GO - Cellular Component
GO:0016459 : myosin complex (https://www.ebi.ac.uk/QuickGO/term/GO:0016459)

UniProtKB Homo sapiens
Q9H6N6 (http://www.uniprot.org/uniprot/Q9H6N6)

GenebankID or UniProtKB
AC004834 (https://www.ncbi.nlm.nih.gov/nuccore/AC004834)

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

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

Deletion Size

1-9 bp

Molecular Details of the Mutation

2bp deletion at codon 660

Experimental Evidence

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

Main Reference

Myosin gene mutation correlates with anatomical changes in the human lineage. (2004) (https://pubmed.ncbi.nlm.nih.gov/15042088)

Authors

Stedman HH; Kozyak BW; Nelson A; Thesier DM; Su LT; Low DW; Bridges CR; Shrager JB; Minugh-Purvis N; Mitchell MA

Abstract

Powerful masticatory muscles are found in most primates, including chimpanzees and gorillas, and were part of a prominent adaptation of Australopithecus and Paranthropus, extinct genera of the family Hominidae. In contrast, masticatory muscles are considerably smaller in both modern and fossil members of Homo. The evolving hominid masticatory apparatus--traceable to a Late Miocene, chimpanzee-like morphology--shifted towards a pattern of gracilization nearly simultaneously with accelerated encephalization in early Homo. Here, we show that the gene encoding the predominant myosin heavy chain (MYH) expressed in these muscles was inactivated by a frameshifting mutation after the lineages leading to humans and chimpanzees diverged. Loss of this protein isoform is associated with marked size reductions in individual muscle fibres and entire masticatory muscles. Using the coding sequence for the myosin rod domains as a molecular clock, we estimate that this mutation appeared approximately 2.4 million years ago, predating the appearance of modern human body size and emigration of Homo from Africa. This represents the first proteomic distinction between humans and chimpanzees that can be correlated with a traceable anatomic imprint in the fossil record.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS