GEPHE SUMMARY

Gephebase Gene

Prdm9 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Prdm9^#gephebase-summary-title)

Entry Status

Published

GepheID

GP00000923

Main curator

Martin

PHENOTYPIC CHANGE

Trait Category

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

Trait

Recombination rate (use of recombination hotspots) (https://www.gephebase.org/search-criteria?/and+Trait=^Recombination rate (use of recombination hotspots)^#gephebase-summary-title)

Trait State in Taxon A

Homo sapiens

Trait State in Taxon B

Homo sapiens

Ancestral State

Data not curated

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

Prdm9

Synonyms

Hst1; Meisetz

String

10090.ENSMUSP00000131871 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000131871)

Sequence Similarities

Belongs to the class V-like SAM-binding methyltransferase superfamily.

GO - Molecular Function

GO:0000977 : RNA polymerase II regulatory region sequence-specific DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0000977)
GO:0043565 : sequence-specific DNA binding

UniProtKB Mus musculus

Q96EQ9 (http://www.uniprot.org/uniprot/Q96EQ9)

GenebankID or UniProtKB

ADA68172 (https://www.ncbi.nlm.nih.gov/nuccore/ADA68172)

(https://www.ebi.ac.uk/QuickGO/term/GO:0043565)
GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)
GO:0044212 : transcription regulatory region DNA binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0044212)
GO:0031490 : chromatin DNA binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0031490)
GO:0042800 : histone methyltransferase activity (H3-K4 specific)
(https://www.ebi.ac.uk/QuickGO/term/GO:0042800)
GO:0018024 : histone-lysine N-methyltransferase activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0018024)
GO:0010844 : recombination hotspot binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0010844)

GO - Biological Process

GO:0045944 : positive regulation of transcription by RNA polymerase II
(https://www.ebi.ac.uk/QuickGO/term/GO:0045944)
GO:0007283 : spermatogenesis (https://www.ebi.ac.uk/QuickGO/term/GO:0007283)
GO:0016584 : nucleosome positioning
(https://www.ebi.ac.uk/QuickGO/term/GO:0016584)
GO:0034968 : histone lysine methylation
(https://www.ebi.ac.uk/QuickGO/term/GO:0034968)
GO:0016571 : histone methylation (https://www.ebi.ac.uk/QuickGO/term/GO:0016571)
GO:0006311 : meiotic gene conversion
(https://www.ebi.ac.uk/QuickGO/term/GO:0006311)
GO:0060903 : positive regulation of meiosis I
(https://www.ebi.ac.uk/QuickGO/term/GO:0060903)
GO:0010845 : positive regulation of reciprocal meiotic recombination
(https://www.ebi.ac.uk/QuickGO/term/GO:0010845)

GO - Cellular Component

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:0005694 : chromosome (https://www.ebi.ac.uk/QuickGO/term/GO:0005694)

Presumptive Null

Unknown (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Unknown^#gephebase-summary-title)

Molecular Type

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

Aberration Type

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

Molecular Details of the Mutation

Various haplotypes

Experimental Evidence

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

Main Reference

Variants of the protein PRDM9 differentially regulate a set of human meiotic recombination hotspots highly active in African populations. (2011) (https://pubmed.ncbi.nlm.nih.gov/21750151)

Authors

Berg IL; Neumann R; Sarbajna S; Odenthal-Hesse L; Butler NJ; Jeffreys AJ

Abstract

PRDM9 is a major specifier of human meiotic recombination hotspots, probably via binding of its zinc-finger repeat array to a DNA sequence motif associated with hotspots. However, our view of PRDM9 regulation, in terms of motifs defined and hotspots studied, has a strong bias toward the PRDM9 A variant particularly common in Europeans. We show that population diversity can reveal a second class of hotspots specifically activated by PRDM9 variants common in Africans but rare in Europeans. These African-enhanced hotspots nevertheless share very similar properties with their counterparts activated by the A variant. The specificity of hotspot activation is such that individuals with differing PRDM9 genotypes, even within the same population, can use substantially if not completely different sets of hotspots. Each African-enhanced hotspot is activated by a distinct spectrum of PRDM9 variants, despite the fact that all are predicted to bind the same sequence motif. This differential activation points to complex interactions between the zinc-finger array and hotspots and identifies features of the array that might be important in controlling hotspot activity.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS