GEPHE SUMMARY

Gephebase Gene
Prdm9

GephelD
GP00000923

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Physiology

Trait
Recombination rate (use of recombination hotspots)

Trait State in Taxon A
Homo sapiens

Trait State in Taxon B
Homo sapiens

Ancestral State
Data not curated

Taxonomic Status
Intraspecific

Taxon A

Latin Name
Homo sapiens

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)

NCBI Taxonomy ID
9606

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Homo sapiens

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)

NCBI Taxonomy ID
9606

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
Prdm9

Synonyms
Hst1; Meisetz

String
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
GO:0043565 : sequence-specific DNA binding
GO:0046872 : metal ion binding
GO:0044212 : transcription regulatory region DNA binding
GO:0031490 : chromatin DNA binding
GO:0042800 : histone methyltransferase activity (H3-K4 specific)
GO:0018024 : histone-lysine N-methyltransferase activity
GO:0010844 : recombination hotspot binding

UniProtKB Mus musculus
Q96EQ9

GenebankID or UniProtKB
ADA68172

GO - Biological Process
GO:0045944 : positive regulation of transcription by RNA polymerase II
GO:0007283 : spermatogenesis
GO:0016584 : nucleosome positioning
GO:0034968 : histone lysine methylation
GO:0016571 : histone methylation
GO:0006311 : meiotic gene conversion
GO:0060903 : positive regulation of meiosis I
GO:0010845 : positive regulation of reciprocal meiotic recombination

GO - Cellular Component
GO:0005654 : nucleoplasm
GO:0005634 : nucleus
GO:0005694 : chromosome

Presumptive Null
Unknown

Molecular Type
Unknown

Aberration Type
Unknown

Molecular Details of the Mutation
Various haplotypes

Experimental Evidence
Candidate Gene

Main Reference
Variants of the protein PRDM9 differentially regulate a set of human meiotic recombination hotspots highly active in African populations. (2011)

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