GEPHE SUMMARY

Gephebase Gene

CYP1A2 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^CYP1A2^#gephebase-summary-title)

Entry Status

Published

GepheID

GP00000199

Main curator

Martin

PHENOTYPIC CHANGE

Trait Category

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

Trait

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

Trait State in Taxon A

Homo sapiens

Trait State in Taxon B

Homo sapiens - Japanese

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

CYP1A2

Synonyms

CP12; CYPIA2; P3-450; P450(PA)

String

9606.ENSP00000342007 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000342007)

Sequence Similarities

Belongs to the cytochrome P450 family.

GO - Molecular Function

GO:0016491 : oxidoreductase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0016491)
GO:0019899 : enzyme binding (https://www.ebi.ac.uk/QuickGO/term/GO:0019899)
GO:0020037 : heme binding (https://www.ebi.ac.uk/QuickGO/term/GO:0020037)
GO:0005506 : iron ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005506)

UniProtKB Homo sapiens

P05177 (http://www.uniprot.org/uniprot/P05177)

GenebankID or UniProtKB

BC067424 (https://www.ncbi.nlm.nih.gov/nuccore/BC067424)

GO:0004497 : monooxygenase activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0004497)
GO:0070330 : aromatase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0070330)
GO:0034875 : caffeine oxidase activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0034875)
GO:0032451 : demethylase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0032451)
GO:0009055 : electron transfer activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0009055)
GO:0016712 : oxidoreductase activity, acting on paired donors, with incorporation or reduction of molecular oxygen, reduced flavin or flavoprotein as one donor, and incorporation of one atom of oxygen (https://www.ebi.ac.uk/QuickGO/term/GO:0016712)

GO - Biological Process

GO:0055114 : oxidation-reduction process
(https://www.ebi.ac.uk/QuickGO/term/GO:0055114)
GO:0032259 : methylation (https://www.ebi.ac.uk/QuickGO/term/GO:0032259)
GO:0032496 : response to lipopolysaccharide
(https://www.ebi.ac.uk/QuickGO/term/GO:0032496)
GO:0009820 : alkaloid metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0009820)
GO:0045333 : cellular respiration (https://www.ebi.ac.uk/QuickGO/term/GO:0045333)
GO:0071276 : cellular response to cadmium ion
(https://www.ebi.ac.uk/QuickGO/term/GO:0071276)
GO:0071280 : cellular response to copper ion
(https://www.ebi.ac.uk/QuickGO/term/GO:0071280)
GO:0018894 : dibenzo-p-dioxin metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0018894)
GO:0042737 : drug catabolic process (https://www.ebi.ac.uk/QuickGO/term/GO:0042737)
GO:0017144 : drug metabolic process (https://www.ebi.ac.uk/QuickGO/term/GO:0017144)
GO:0019373 : epoxygenase P450 pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0019373)
GO:0042738 : exogenous drug catabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0042738)
GO:0046483 : heterocycle metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0046483)
GO:0050665 : hydrogen peroxide biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0050665)
GO:0042759 : long-chain fatty acid biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0042759)
GO:0030324 : lung development (https://www.ebi.ac.uk/QuickGO/term/GO:0030324)
GO:0032787 : monocarboxylic acid metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0032787)
GO:0016098 : monoterpenoid metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0016098)
GO:0097267 : omega-hydroxylase P450 pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0097267)
GO:0071615 : oxidative deethylation (https://www.ebi.ac.uk/QuickGO/term/GO:0071615)
GO:0070989 : oxidative demethylation
(https://www.ebi.ac.uk/QuickGO/term/GO:0070989)
GO:0006778 : porphyrin-containing compound metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006778)
GO:0009791 : post-embryonic development
(https://www.ebi.ac.uk/QuickGO/term/GO:0009791)
GO:0010468 : regulation of gene expression
(https://www.ebi.ac.uk/QuickGO/term/GO:0010468)
GO:0032355 : response to estradiol (https://www.ebi.ac.uk/QuickGO/term/GO:0032355)
GO:0035902 : response to immobilization stress
(https://www.ebi.ac.uk/QuickGO/term/GO:0035902)
GO:0006706 : steroid catabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006706)
GO:0009403 : toxin biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0009403)
GO:0006805 : xenobiotic metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006805)

GO - Cellular Component

GO:0043231 : intracellular membrane-bounded organelle
(https://www.ebi.ac.uk/QuickGO/term/GO:0043231)
GO:0005789 : endoplasmic reticulum membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0005789)
GO:0031090 : organelle membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0031090)

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

F186L

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

Six novel nonsynonymous CYP1A2 gene polymorphisms: catalytic activities of the naturally occurring variant enzymes. (2004) (https://pubmed.ncbi.nlm.nih.gov/14563787)

Authors

Murayama N; Soyama A; Saito Y; Nakajima Y; Komamura K; Ueno K; Kamakura S; Kitakaze M; Kimura H; Goto Y; Saitoh O; Katoh M; Ohnuma T; Kawai M; Sugai K; Ohtsuki T; Suzuki C; Minami N; Ozawa S; Sawada J

Abstract

Six novel nonsynonymous nucleotide alterations were found in the cytochrome P450 1A2 gene in a Japanese population, which resulted in the following amino acid substitutions: T83M, E168Q, F186L, S212C, G299A, and T438I. These individuals were heterozygous for the amino acid substitutions. The potential functional alterations caused by the amino acid substitutions were characterized by a cDNA-mediated expression system using Chinese hamster V79 cells. Among the six CYP1A2 variants, F186L showed the most profound and statistically significant reduction in O-deethylation of phenacetin and 7-ethoxyresorufin. Kinetic analyses performed for the ethoxyresorufin O-deethylation revealed that the Vmax of the F186L variant was approximately 5% of that of the CYP1A2 wild type, despite a 5-fold lower Km value of the variant, the consequence of which was reduced enzymatic activity toward the substrate. Thus, for the first time, phenylalanine at residue 186 is suggested to be a critical amino acid for catalytic activity.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS