

## GEPHE SUMMARY

**Gephebase Gene**  
CYP1A2

**Entry Status**  
Published

**GepheID**  
GP00000199

**Main curator**  
Martin

## PHENOTYPIC CHANGE

**Trait Category**  
Physiology

**Trait**  
Enzymatic activity

**Trait State in Taxon A**  
Homo sapiens

**Trait State in Taxon B**  
Homo sapiens - Japanese

**Ancestral State**  
Taxon A

**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; Haplorhini; Simiiformes; Catarrhini; Hominoidea; Hominidae; Homininae; Homo

**Parent**  
Homo () - (Rank: genus)

**NCBI Taxonomy ID**  
9606

**is Taxon A an Intraspecies?**  
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; Haplorhini; Simiiformes; Catarrhini; Hominoidea; Hominidae; Homininae; Homo

**Parent**  
Homo () - (Rank: genus)

**NCBI Taxonomy ID**  
9606

**is Taxon B an Intraspecies?**  
No

## GENOTYPIC CHANGE

**Generic Gene Name**  
CYP1A2

**Synonyms**  
CP12; CYP1A2; P3-450; P450(PA)

**String**  
9606.ENSP00000342007

**Sequence Similarities**  
Belongs to the cytochrome P450 family.

**GO - Molecular Function**  
GO:0016491 : oxidoreductase activity  
GO:0019899 : enzyme binding  
GO:0020037 : heme binding  
GO:0005506 : iron ion binding  
GO:0004497 : monooxygenase activity  
GO:0070330 : aromatase activity  
GO:0034875 : caffeine oxidase activity  
GO:0032451 : demethylase activity

**UniProtKB Homo sapiens**  
P05177

**GenebankID or UniProtKB**  
BC067424

GO:0009055 : electron transfer activity  
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

#### GO - Biological Process

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

#### GO - Cellular Component

GO:0043231 : intracellular membrane-bounded organelle  
GO:0005789 : endoplasmic reticulum membrane  
GO:0031090 : organelle membrane

#### Presumptive Null

No

#### Molecular Type

Coding

#### Aberration Type

SNP

#### SNP Coding Change

Nonsynonymous

#### Molecular Details of the Mutation

F186L

#### Experimental Evidence

Candidate Gene

	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)

#### 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 Genes](#)

No matches found.

[Related Haplotypes](#)

No matches found.

**EXTERNAL LINKS**

**COMMENTS**