

Human NAT2 alleles (Haplotypes) (updated September 29, 2009)

<u>NAT2 Allele (Haplotype)^a</u>	<u>Nucleotide Change(s) and rs Identifiers^b</u>	<u>Amino Acid Change(s)^b</u>	<u>Phenotype^c</u>	<u>References</u>
<i>NAT2*4</i>	Reference	Reference	Rapid	1-17
<i>NAT2*5A</i>	341T>C (rs1801280) 481C>T (rs1799929)	I114T L161L (synonymous)	Slow	2,9,10,18,19
<i>NAT2*5B</i>	341T>C (rs1801280) 481C>T (rs1799929) 803A>G (rs1208)	I114T L161L (synonymous) K268R	Slow	3, 9-17, 19,21
<i>NAT2*5C</i>	341T>C (rs1801280) 803A>G (rs1208)	I114T K268R	Slow	9,10,18-20,22
<i>NAT2*5D</i>	341T>C (rs1801280)	I114T	Slow	12,15,17,23-26
<i>NAT2*5E</i>	341T>C (rs1801280) 590G>A (rs1799930)	I114T R197Q	Slow	23
<i>NAT2*5F</i>	341T>C (rs1801280) 481C>T (rs1799929) 759C>T 803A>G (rs1208)	I114T L161L (synonymous) V253V (synonymous) K268R	Slow	27
<i>NAT2*5G</i>	282C>T (rs1041983) 341T>C (rs1801280) 481C>T (rs1799929) 803A>G (rs1208)	Y94Y (synonymous) I114T L161L (synonymous) K268R	Slow	28
<i>NAT2*5H</i>	341T>C (rs1801280) 481C>T (rs1799929) 803A>G (rs1208) 859Del	I114T L161L (synonymous) K268R S287 Frameshift	Slow	28
<i>NAT2*5I</i>	341T>C (rs1801280) 411A>T (rs4986997) 481C>T (rs1799929) 803A>G (rs1208)	I114T L137F L161L (synonymous) K268R	Slow	16
<i>NAT2*5J^d</i>	282C>T (rs1041983) 341T>C (rs1801280) 590G>A (rs1799930)	Y94Y (synonymous) I114T R197Q	Slow	29

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<i>NAT2*5K</i>	282C>T (rs1041983) 341T>C (rs1801280)	Y94Y (synonymous) I114T		30
<i>NAT2*5L</i>	70T>A 341T>C (rs1801280) 481C>T (rs1799929) 803A>G (rs1208)	L24I I114T L161L (synonymous) K268R		30,31
<i>NAT2*5M</i>	341T>C (rs1801280) 481C>T (rs1799929) 803A>G (rs1208) 838G>A	I114T L161L (synonymous) K268R V280M		32
<i>NAT2*5N</i>	341T>C (rs1801280) 472A>C 481C>T (rs1799929) 803A>G (rs1208)	I114T I158L L161L (synonymous) K268R		53
<i>NAT2*5O</i>	203G>A (rs72466458) 341T>C (rs1801280) 481C>T (rs1799929) 803A>G (rs1208)	C68Y I114T L161L (synonymous) K268R		53
<i>NAT2*6A</i>	282C>T (rs1041983) 590G>A (rs1799930)	Y94Y (synonymous) R197Q	Slow	1-3,9-17,19,22
<i>NAT2*6B</i>	590G>A (rs1799930)	R197Q	Slow	12,15,17,19,24,25
<i>NAT2*6C</i>	282C>T (rs1041983) 590G>A (rs1799930) 803A>G (rs1208)	Y94Y (synonymous) R197Q K268R	Slow	24,25,30,33
<i>NAT2*6D</i>	111T>C 282C>T (rs1041983) 590G>A (rs1799930)	F37F (synonymous) Y94Y (synonymous) R197Q	Slow	26
<i>NAT2*6E</i>	481C>T (rs1799929) 590G>A (rs1799930)	L161L (synonymous) R197Q	Slow	34
<i>NAT2*6F</i>	590G>A (rs1799930) 803A>G (rs1208)	R197Q K268R		31

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<i>NAT2*6G</i>	282C>T (rs1041983) 518A>G 590G>A (rs1799930)	Y94Y (synonymous) K173R R197Q		31
<i>NAT2*6H</i>	282C>T (rs1041983) 590G>A (rs1799930) 766A>G	Y94Y (synonymous) R197Q K256E		31,53
<i>NAT2*6I</i>	282C>T (rs1041983) 590G>A (rs1799930) 838G>A 857G>A (rs1799931)	Y94Y (synonymous) R197Q V280M G286E		31
<i>NAT2*6J</i>	282C>T (rs1041983) 590G>A (rs1799930) 857G>A (rs1799931)	Y94Y (synonymous) R197Q G286E		32
<i>NAT2*6K</i>	282C>T (rs1041983) 590G>A (rs1799930) 638C>T	Y94Y (synonymous) R197Q P213L		32
<i>NAT2*6L</i>	282C>T (rs1041983) 345C>T 590G>A (rs1799930)	Y94Y (synonymous) D115D (synonymous) R197Q		32
<i>NAT2*6M</i>	152G>T (rs72466457) 282C>T (rs1041983) 590G>A (rs1799930)	Gly51Val Y94Y (synonymous) R197Q		53
<i>NAT2*7A</i>	857G>A (rs1799931)	G286E	Slow Substrate dependent?	2,11,12,15,17,19,33
<i>NAT2*7B</i>	282C>T (rs1041983) 857G>A (rs1799931)	Y94Y (synonymous) G286E	Slow Substrate dependent?	1,9-18, 33

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<i>NAT2*10</i>	499G>A	E167K	Slow Substrate dependent? ^f	11,15-17
<i>NAT2*11A</i>	481C>T (rs1799929)	L161L (synonymous)	Rapid	12,15,17,35
<i>NAT2*11B</i>	481C>T (rs1799929) 859Del	L161L (synonymous) S287 Frameshift	Unknown	28
<i>NAT2*12A</i>	803A>G (rs1208)	K268R	Rapid	12,15,17,19,36,37
<i>NAT2*12B</i>	282C>T (rs1041983) 803A>G (rs1208)	Y94Y (synonymous) K268R	Rapid	9,10,19,25
<i>NAT2*12C</i>	481C>T (rs1799929) 803A>G (rs1208)	L161L (synonymous) K268R	Rapid	9,10,20,24,25,37
<i>NAT2*12D</i>	364G>A (rs4986996) 803A>G (rs1208)	D122N K268R	Slow	16
<i>NAT2*12E</i>	282C>T (rs1041983) 578C>T 803A>G (rs1208)	Y94Y (synonymous) T193M K268R		30,31,53
<i>NAT2*12F</i>	622T>C 803A>G (rs1208)	Y208H K268R		30,53
<i>NAT2*12G</i>	609G>T 803A>G (rs1208)	E203D K268R		31
<i>NAT2*12H</i>	403C>G 803A>G (rs1208)	L135V K268R		32
<i>NAT2*12I</i>	228C>T (rs72466459) 803A>G (rs1208)	T76T (synonymous) K268R		53
<i>NAT2*12J</i>	29C>T (rs72466456) 803A>G (rs1208)	I10T K268R		53

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<i>NAT2*13A</i>	282C>T (rs1041983)	Y94Y (synonymous)	Rapid	9,10,12-15, 17,19,22,23,37,38
<i>NAT2*13B</i>	282C>T (rs1041983) 578C>T	Y94Y (synonymous) T193M		31
<i>NAT2*14A</i>	191G>A (rs1801279)	R64Q	Slow	9,10,12- 15,17,20,39
<i>NAT2*14B</i>	191G>A (rs1801279) 282C>T (rs1041983)	R64Q Y94Y (synonymous)	Slow	39,40
<i>NAT2*14C</i>	191G>A (rs1801279) 341T>C (rs1801280) 481C>T (rs1799929) 803A>G (rs1208)	R64Q I114T L161L (synonymous) K268R	Slow	23-25,33
<i>NAT2*14D</i>	191G>A (rs1801279) 282C>T (rs1041983) 590G>A (rs1799930)	R64Q Y94Y (synonymous) R197Q	Slow	23,25,33
<i>NAT2*14E</i>	191G>A (rs1801279) 803A>G (rs1208)	R64Q K268R	Slow	23,31,33
<i>NAT2*14F</i>	191G>A (rs1801279) 341T>C (rs1801280) 803A>G (rs1208)	R64Q I114T K268R	Slow	25
<i>NAT2*14G</i>	191G>A (rs1801279) 282C>T (rs1041983) 803A>G (rs1208)	R64Q Y94Y (synonymous) K268R	Slow	26
<i>NAT2*14H</i>	191G>A (rs1801279) 282C>T (rs1041983) 683C>T	R64Q Y94Y (synonymous) P228L		31
<i>NAT2*14I</i>	191G>A (rs1801279) 481C>T (rs1799929) 803A>G (rs1208)	R64Q L161L (synonymous) K268R		52
<i>NAT2*17</i>	434A>C	Q145P	Slow	9,10,12,15,41

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<i>NAT2*18</i>	845A>C	K282T	Rapid	9,10,12,15,41
<i>NAT2*19</i>	190C>T (rs1805158)	R64W	Slow	42-44
<i>NAT2*20</i>	600A>G(rs72466461)	E200E(synonymous)		53
<i>NAT2*21</i>	458C>T(rs72466460)	T153I		53

Notes:

- ^aHuman *NAT2* alleles should be written in upper case and italicized. Protein products of the alleles are also upper case but not italicized and the asterisk is omitted. For example, the allele *NAT2*4* encodes the protein NAT2 4.
- ^aReference gene sequence published in [Genbank Assession Number X14672](#). The frequency of individual SNPs and alleles in various ethnic groups is available online from the National Cancer Institute's [SNP500Cancer database](#).
- ^a*NAT2*4* has historically been designated "wildtype". Since it is the most common occurring alleles in some but not all ethnic groups the designation of "wildtype" allele is somewhat arbitrary and is dependent upon the ethnicity of the population studied.
- ^bSignature SNP for cluster is shown in red font.
- ^cPhenotype assignments reflect most current research but are not necessarily a consensus of all research papers. Evidence also exists for heterogeneity within the "slow" acetylator phenotype. For comprehensive reviews, see references 45-49.
- ^dThe SNPs on *NAT2*5J* were determined by RFLP methods which are indirect.
- Although additional SNPs have been identified outside the open reading frame (e.g., see refs 31,32,50), they will not be named until a functional effect is observed. SNPs should be identified by designating "A" of the ATG translation initiation codon as number 1. SNPs upstream of this site are designated by negative numbers and SNPs downstream of this site are designated by positive numbers (51).

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