

## Human NAT2 alleles (Haplotypes) (updated July 22, 2011)

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

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<i>NAT2*5K</i>	<b>282C&gt;T</b> (rs1041983) <b>341T&gt;C</b> (rs1801280)	<b>Y94Y</b> (synonymous) <b>I114T</b>		<b>30</b>
<i>NAT2*5L</i>	<b>70T&gt;A</b> <b>341T&gt;C</b> (rs1801280) <b>481C&gt;T</b> (rs1799929) <b>803A&gt;G</b> (rs1208)	<b>L24I</b> <b>I114T</b> <b>L161L</b> (synonymous) <b>K268R</b>		<b>30,31</b>
<i>NAT2*5M</i>	<b>341T&gt;C</b> (rs1801280) <b>481C&gt;T</b> (rs1799929) <b>803A&gt;G</b> (rs1208) <b>838G&gt;A</b>	<b>I114T</b> <b>L161L</b> (synonymous) <b>K268R</b> <b>V280M</b>		<b>32</b>
<i>NAT2*5N</i>	<b>341T&gt;C</b> (rs1801280) <b>472A&gt;C</b> <b>481C&gt;T</b> (rs1799929) <b>803A&gt;G</b> (rs1208)	<b>I114T</b> <b>I158L</b> <b>L161L</b> (synonymous) <b>K268R</b>		<b>53</b>
<i>NAT2*5O</i>	<b>203G&gt;A</b> (rs72466458) <b>341T&gt;C</b> (rs1801280) <b>481C&gt;T</b> (rs1799929) <b>803A&gt;G</b> (rs1208)	<b>C68Y</b> <b>I114T</b> <b>L161L</b> (synonymous) <b>K268R</b>		<b>53</b>
<i>NAT2*5P</i>	<b>282C&gt;T</b> (rs1041983) <b>341T&gt;C</b> (rs1801280) <b>481C&gt;T</b> (rs1799929) <b>578C&gt;T</b> <b>590G&gt;A</b> (rs1799930) <b>803A&gt;G</b> (rs1208)	<b>Y94Y</b> (synonymous) <b>I114T</b> <b>L161L</b> (synonymous) <b>T193M</b> <b>R197Q</b> <b>K268R</b>		<b>54</b>
<i>NAT2*6A</i>	<b>282C&gt;T</b> (rs1041983) <b>590G&gt;A</b> (rs1799930)	<b>Y94Y</b> (synonymous) <b>R197Q</b>	<b>Slow</b>	<b>1-3,9-17,19,22</b>
<i>NAT2*6B</i>	<b>590G&gt;A</b> (rs1799930)	<b>R197Q</b>	<b>Slow</b>	<b>12,15,17,19,24,25</b>
<i>NAT2*6C</i>	<b>282C&gt;T</b> (rs1041983) <b>590G&gt;A</b> (rs1799930) <b>803A&gt;G</b> (rs1208)	<b>Y94Y</b> (synonymous) <b>R197Q</b> <b>K268R</b>	<b>Slow</b>	<b>24,25,30,33</b>
<i>NAT2*6D</i>	<b>111T&gt;C</b> <b>282C&gt;T</b> (rs1041983) <b>590G&gt;A</b> (rs1799930)	<b>F37F</b> (synonymous) <b>Y94Y</b> (synonymous) <b>R197Q</b>	<b>Slow</b>	<b>26</b>

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<i>NAT2*6E</i>	<b>481C&gt;T</b> (rs1799929) <b>590G&gt;A</b> (rs1799930)	<b>L161L</b> (synonymous) <b>R197Q</b>	<b>Slow</b>	<b>34</b>
<i>NAT2*6F</i>	<b>590G&gt;A</b> (rs1799930) <b>803A&gt;G</b> (rs1208)	<b>R197Q</b> <b>K268R</b>		<b>31</b>
<i>NAT2*6G</i>	<b>282C&gt;T</b> (rs1041983) <b>518A&gt;G</b> <b>590G&gt;A</b> (rs1799930)	<b>Y94Y</b> (synonymous) <b>K173R</b> <b>R197Q</b>		<b>31</b>
<i>NAT2*6H</i>	<b>282C&gt;T</b> (rs1041983) <b>590G&gt;A</b> (rs1799930) <b>766A&gt;G</b>	<b>Y94Y</b> (synonymous) <b>R197Q</b> <b>K256E</b>		<b>31,53</b>
<i>NAT2*6I</i>	<b>282C&gt;T</b> (rs1041983) <b>590G&gt;A</b> (rs1799930) <b>838G&gt;A</b> <b>857G&gt;A</b> (rs1799931)	<b>Y94Y</b> (synonymous) <b>R197Q</b> <b>V280M</b> <b>G286E</b>		<b>31</b>
<i>NAT2*6J</i>	<b>282C&gt;T</b> (rs1041983) <b>590G&gt;A</b> (rs1799930) <b>857G&gt;A</b> (rs1799931)	<b>Y94Y</b> (synonymous) <b>R197Q</b> <b>G286E</b>		<b>32</b>
<i>NAT2*6K</i>	<b>282C&gt;T</b> (rs1041983) <b>590G&gt;A</b> (rs1799930) <b>638C&gt;T</b>	<b>Y94Y</b> (synonymous) <b>R197Q</b> <b>P213L</b>		<b>32</b>
<i>NAT2*6L</i>	<b>282C&gt;T</b> (rs1041983) <b>345C&gt;T</b> <b>590G&gt;A</b> (rs1799930)	<b>Y94Y</b> (synonymous) <b>D115D</b> (synonymous) <b>R197Q</b>		<b>32</b>
<i>NAT2*6M</i>	<b>152G&gt;T</b> (rs72466457) <b>282C&gt;T</b> (rs1041983) <b>590G&gt;A</b> (rs1799930)	<b>Gly51Val</b> <b>Y94Y</b> (synonymous) <b>R197Q</b>		<b>53</b>
<i>NAT2*6N</i>	<b>282C&gt;T</b> (rs1041983) <b>481C&gt;T</b> (rs1799929) <b>590G&gt;A</b> (rs1799930)	<b>Y94Y</b> (synonymous) <b>L161L</b> (synonymous) <b>R197Q</b>		<b>55</b>
<i>NAT2*6O<sup>e</sup></i>	<b>282C&gt;T</b> (rs1041983) <b>481C&gt;T</b> (rs1799929) <b>838G&gt;A</b>	<b>Y94Y</b> (synonymous) <b>L161L</b> (synonymous) <b>V280M</b>		<b>56</b>

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<i>NAT2*7A</i>	<b>857G&gt;A</b> (rs1799931)	<b>G286E</b>	Slow Substrate dependent?	2,11,12,15,17,19,33
<i>NAT2*7B</i>	<b>282C&gt;T</b> (rs1041983) <b>857G&gt;A</b> (rs1799931)	<b>Y94Y</b> (synonymous) <b>G286E</b>	Slow Substrate dependent?	1,9-18, 33
<i>NAT2*7C</i>	<b>282C&gt;T</b> (rs1041983) <b>803A&gt;G</b> (rs1208) <b>857G&gt;A</b> (rs1799931)	<b>Y94Y</b> (synonymous) <b>K268R</b> <b>G286E</b>		54
<i>NAT2*7D</i>	<b>191G&gt;A</b> (rs1801279) <b>282C&gt;T</b> (rs1041983) <b>857G&gt;A</b> (rs1799931)	<b>R64Q</b> <b>Y94Y</b> (synonymous) <b>G286E</b>		53
<i>NAT2*7E<sup>e</sup></i>	<b>282C&gt;T</b> (rs1041983) <b>481C&gt;T</b> (rs1799929) <b>857G&gt;A</b> (rs1799931)	<b>Y94Y</b> (synonymous) <b>L161L</b> (synonymous) <b>G286E</b>		56
<i>NAT2*10</i>	<b>499G&gt;A</b>	<b>E167K</b>	Slow Substrate dependent? <sup>f</sup>	11,15-17
<i>NAT2*11A</i>	<b>481C&gt;T</b> (rs1799929)	<b>L161L</b> (synonymous)	Rapid	12,15,17,35
<i>NAT2*11B</i>	<b>481C&gt;T</b> (rs1799929) <b>859Del</b>	<b>L161L</b> (synonymous) <b>S287 Frameshift</b>	Unknown	28
<i>NAT2*12A</i>	<b>803A&gt;G</b> (rs1208)	<b>K268R</b>	Rapid	12,15,17,19,36,37
<i>NAT2*12B</i>	<b>282C&gt;T</b> (rs1041983) <b>803A&gt;G</b> (rs1208)	<b>Y94Y</b> (synonymous) <b>K268R</b>	Rapid	9,10,19,25
<i>NAT2*12C</i>	<b>481C&gt;T</b> (rs1799929) <b>803A&gt;G</b> (rs1208)	<b>L161L</b> (synonymous) <b>K268R</b>	Rapid	9,10,20,24,25,37
<i>NAT2*12D</i>	<b>364G&gt;A</b> (rs4986996) <b>803A&gt;G</b> (rs1208)	<b>D122N</b> <b>K268R</b>	Slow	16

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<i>NAT2*12E</i>	<b>282C&gt;T</b> (rs1041983) <b>578C&gt;T</b> <b>803A&gt;G</b> (rs1208)	<b>Y94Y</b> (synonymous) <b>T193M</b> <b>K268R</b>		<b>30,31,53</b>
<i>NAT2*12F</i>	<b>622T&gt;C</b> <b>803A&gt;G</b> (rs1208)	<b>Y208H</b> <b>K268R</b>		<b>30,53</b>
<i>NAT2*12G</i>	<b>609G&gt;T</b> <b>803A&gt;G</b> (rs1208)	<b>E203D</b> <b>K268R</b>		<b>31</b>
<i>NAT2*12H</i>	<b>403C&gt;G</b> <b>803A&gt;G</b> (rs1208)	<b>L135V</b> <b>K268R</b>		<b>32</b>
<i>NAT2*12I</i>	<b>228C&gt;T</b> (rs72466459) <b>803A&gt;G</b> (rs1208)	synonymous <b>K268R</b>		<b>53</b>
<i>NAT2*12J</i>	<b>29C&gt;T</b> (rs72466456) <b>803A&gt;G</b> (rs1208)	<b>I10T</b> <b>K268R</b>		<b>53</b>
<i>NAT2*13A</i>	<b>282C&gt;T</b> (rs1041983)	<b>Y94Y</b> (synonymous)	<b>Rapid</b>	<b>9,10,12-15, 17,19,22,23,37,38</b>
<i>NAT2*13B</i>	<b>282C&gt;T</b> (rs1041983) <b>578C&gt;T</b>	<b>Y94Y</b> (synonymous) <b>T193M</b>		<b>31</b>
<i>NAT2*14A</i>	<b>191G&gt;A</b> (rs1801279)	<b>R64Q</b>	<b>Slow</b>	<b>9,10,12- 15,17,20,39</b>
<i>NAT2*14B</i>	<b>191G&gt;A</b> (rs1801279) <b>282C&gt;T</b> (rs1041983)	<b>R64Q</b> <b>Y94Y</b> (synonymous)	<b>Slow</b>	<b>39,40</b>
<i>NAT2*14C</i>	<b>191G&gt;A</b> (rs1801279) <b>341T&gt;C</b> (rs1801280) <b>481C&gt;T</b> (rs1799929) <b>803A&gt;G</b> (rs1208)	<b>R64Q</b> <b>I114T</b> <b>L161L</b> (synonymous) <b>K268R</b>	<b>Slow</b>	<b>23-25,33</b>
<i>NAT2*14D</i>	<b>191G&gt;A</b> (rs1801279) <b>282C&gt;T</b> (rs1041983) <b>590G&gt;A</b> (rs1799930)	<b>R64Q</b> <b>Y94Y</b> (synonymous) <b>R197Q</b>	<b>Slow</b>	<b>23,25,33</b>
<i>NAT2*14E</i>	<b>191G&gt;A</b> (rs1801279) <b>803A&gt;G</b> (rs1208)	<b>R64Q</b> <b>K268R</b>	<b>Slow</b>	<b>23,31,33</b>

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<i>NAT2*14F</i>	<b>191G&gt;A</b> (rs1801279) <b>341T&gt;C</b> (rs1801280) <b>803A&gt;G</b> (rs1208)	<b>R64Q</b> <b>I114T</b> <b>K268R</b>	Slow	25
<i>NAT2*14G</i>	<b>191G&gt;A</b> (rs1801279) <b>282C&gt;T</b> (rs1041983) <b>803A&gt;G</b> (rs1208)	<b>R64Q</b> <b>Y94Y</b> (synonymous) <b>K268R</b>	Slow	26
<i>NAT2*14H</i>	<b>191G&gt;A</b> (rs1801279) <b>282C&gt;T</b> (rs1041983) <b>683C&gt;T</b>	<b>R64Q</b> <b>Y94Y</b> (synonymous) <b>P228L</b>		31
<i>NAT2*14I</i>	<b>191G&gt;A</b> (rs1801279) <b>481C&gt;T</b> (rs1799929) <b>803A&gt;G</b> (rs1208)	<b>R64Q</b> <b>L161L</b> (synonymous) <b>K268R</b>		52
<i>NAT2*17</i>	<b>434A&gt;C</b>	<b>Q145P</b>	Slow	9,10,12,15,41
<i>NAT2*18</i>	<b>845A&gt;C</b>	<b>K282T</b>	Rapid	9,10,12,15,41
<i>NAT2*19</i>	<b>190C&gt;T</b> (rs1805158)	<b>R64W</b>	Slow	42-44
<i>NAT2*20</i>	<b>600A&gt;G</b> (rs72466461)	synonymous		53
<i>NAT2*21</i>	<b>458C&gt;T</b> (rs72466460)	<b>T153I</b>		53

### Notes:

- <sup>a</sup>Human *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.
- <sup>a</sup>Reference 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](#).
- <sup>a</sup>*NAT2\*4* has historically been designated "wildtype". Since it is the most common occurring allele 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.
- <sup>b</sup>Signature SNP for cluster is shown in red font.

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- <sup>c</sup>Phenotype 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.
- <sup>d</sup>The SNPs on NAT2\*5J were determined by RFLP methods which are indirect.
- <sup>e</sup>Haplotypes predicted from genotypes by computational PHASE analysis.
- 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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