Allele ¹	Major Nucleotide Variation	dbSNP Number ²	Effect on CYP2C19 Protein
*1	-	-	-
*2	c.681G>A	rs4244285	Splicing defect
*3	c.636G>A	rs4986893	W212X
* 4 ³	c.1A>G	rs28399504	M1V
*5	c.1297C>T	rs56337013	R433W
*6	c.395G>A	rs72552267	R132Q
*7	c.819+2T>A	rs72558186	Splicing defect
*8	c.358T>C	rs41291556	W120R
*174	c806C>T	rs12248560	Increased expression

Supplemental Table S1. Commonly tested *CYP2C19* variant alleles and their effect on CYP2C19 protein

¹ See Human Cytochrome P450 Allele Nomenclature Committee (<u>http://www.cypalleles.ki.se</u>) for comprehensive haplotype definitions of *CYP2C19* variant alleles and updated allele information.

² RefSNP accession ID number (<u>http://www.ncbi.nlm.nih.gov/snp/</u>).

³ Of note, the *CYP2C19*4* loss-of-function allele has been identified in linkage disequilibrium with *17 (c.-806C>T) in certain ethnic subpopulations and this haplotype is designated *CYP2C19*4B* (38).

⁴ There is linkage disequilibrium between c.681G and c.-806T (e.g., |D'|=1.0 and $r^2=0.064$ in CEU HapMap sample; |D'|=1.0 and $r^2=0.065$ in YRI HapMap sample; and |D'|=1.0 and $r^2=0.074$ in CHB HapMap sample). This means that the less common *17 variant (c.-806T) always tracks on the same allele with the more common c.681G. This complicates any interpretation of whether these two variants act independently of one another, and published articles argue both for (29, 35) and against (15, 36, 37) this point.