

Lab Data and Consensus *TMPT* and *NUDT15* Diplotypes*

* Pratt *et al.* Characterization of Reference Materials for *TPMT* and *NUDT15*. A GeT-RM Collaborative Project. *J Mol Diagn* 2022 24:1079-1088

Table 1. Summary of *TPMT* diplotype calls and GeT-RM consensus

| Coriell ID | GeT-RM Consensus | Lab 1 | Lab 2 | Lab 3 | Lab 4 | Lab 5 |
|----------------------|---------------------------|---------|--------|---------|-----------------|-------|
| HG00133 | *1/*2 | *1/*2 | *1/*2 | *1/*2 | NT | NT |
| HG01083 | *1/*2 | *1/*2 | *1/*2 | *1/*2 | NT | *1/*2 |
| NA12753 [^] | *1/*3A[§] | NT | NT | NT | *1/*3A | NT |
| NA15245 [^] | *1/*3A[§] | NT | NT | NT | *1/*3A | NT |
| HG00589 [^] | *1/*3C | NT | NT | NT | *1/*3C | NT |
| NA18855 [^] | *1/*3C | NT | NT | NT | *1/*3C | NT |
| NA18603 | *1/*6 | *1/*6 | *1/*6 | *1/*6 | NT | NT |
| NA19176 [^] | *1/*8 | NT | NT | NT | *1/*8 | NT |
| NA23275 [^] | *1/*8 | NT | NT | NT | *1/*8 | NT |
| NA12751 | *1/*12 | *1/*12 | *1/*12 | *1/*12 | ND | NT |
| HG00276 | *1/*16 | *1/*16 | *1/*16 | *1/*16 | *1/*16 | NT |
| HG00304 | *3A/*16 | *3A/*16 | *3/*16 | *3A/*16 | NT | NT |
| NA12044 | *1/*21 | *1/*21 | *1/*21 | *1/*21 | NT | NT |
| HG02496 | *1/*24 | *1/*24 | *1/*24 | *1/*24 | NT | NT |
| NA10855 | *1/*32 | *1/*32 | *1/*32 | *1/*1 | *1/*32 | NT |
| HG03521 | *1/*46[#] | *1/*33 | *8/*33 | *8/*33 | *1/*46 | NT |
| NA19026 | *8/*33 or *1/*46 | *1/*33 | *8/*33 | *8/*33 | 8/*33 or *1/*46 | NT |
| HG01474 | *1/*40 | *1/*40 | *1/*40 | *1/*40 | NT | NT |

[^]Data from previous GeT-RM study (*J Mol Diagn* 2016, 18:109-123)

§ Phase of heterozygous SNPs was confirmed by 10x Linked-Read Genomics or by trio analysis
 # Novel haplotype; variants identifying *TPMT**8 and *33 were shown to be *in cis* in this sample by trio analysis using 1000 Genomes WGS data
 NT- Sample not tested in study

Table 2. Summary of *NUDT15* diplotype calls and GeT-RM consensus

| Coriell ID | GeT-RM Consensus | Lab 1 | Lab 2 | Lab 3 | Lab 4 | Lab 5 | Lab 6 |
|------------|------------------|-------|-------|-------|-------|---------|-------|
| HG00599 | *2/*2 | *2/*2 | *2/*2 | *2/*2 | *2/*2 | NT | NT |
| NA19079 | *2/*5 | *2/*5 | *2/*5 | NT | *2/*5 | *2/*5^^ | NT |
| NA18992^ | *1/*3 | *1/*3 | NT | NT | *1/*3 | NT | NT |
| HG00524 | *3/*3 | NT | *3/*3 | *3/*3 | *3/*3 | NT | NT |
| NA18564^ | *1/*3 | *1/*3 | NT | NT | *1/*3 | NT | *1/*3 |
| HG01359^ | *1/*4 | NT | NT | *1/*4 | NT | *1/*4 | NT |
| HG01465 | *1/*4 | NT | *1/*4 | *1/*4 | *1/*4 | NT | NT |
| HG00437^ | *1/*5 | NT | NT | *1/*5 | NT | *1/*5 | NT |
| NA23093^ | *1/*5 | *1/*5 | NT | NT | *1/*5 | NT | NT |
| HG00277 | *1/*6 | *1/*6 | *1/*6 | *1/*6 | *1/*6 | NT | NT |
| NA07000 | *1/*6 | *1/*6 | *1/*6 | NT | *1/*6 | NT | NT |
| HG01086 | *2/*9 | *2/*9 | *2/*9 | NT | *2/*9 | *2/*9^^ | NT |

^Previously generated diplotype calls were obtained from laboratories as indicated

^^Confirmed using both targeted genotyping and long-read single molecule real time sequencing (S.A. Scott, personal communication, manuscript in preparation).

NT- Sample not tested in study