

Supplementary Information

Supplementary Table 1. Allele and genotype frequencies of the nine variants genotyped in both the discovery and the replication cohorts. In the replication cohort, DNA was insufficient to genotype the nine variants in all patients; therefore, the number of genotyped patients varies by variant. HWE=Hardy Weinberg Equilibrium; MAF=minor allele frequency

| | | | Discovery Cohort (n=67) | | | Replication Cohort (n=74-104) | | |
|-------------------|---------------------------|-------------------------------|-------------------------|------|------|-------------------------------|------|------|
| Gene | Variant (rs#) | Function | N (%) | MAF | HWE | N (%) | MAF | HWE |
| ABCB1 | IVS9 -44 A>G (rs10276036) | Intronic | | 0.46 | 0.01 | 77 | 0.35 | 0.82 |
| AA | | | 25 (37) | | | 32 (41) | | |
| AG | | | 23 (34) | | | 36 (47) | | |
| GG | | | 19 (28) | | | 9 (12) | | |
| ABCC1 | IVS11 -48 C>T (rs3765129) | Intronic | | 0.17 | 0.08 | 74 | 0.17 | 0.08 |
| CC | | | 48 (72) | | | 49 (66) | | |
| CT | | | 15 (22) | | | 25 (34) | | |
| TT | | | 4 (6) | | | 0 (0) | | |
| ABCC1 | 1684 T>C (rs35605) | Synonymous, Exon 13 | | 0.23 | 0.78 | 95 | 0.22 | 0.03 |
| TT | | | 40 (60) | | | 54 (57) | | |
| TC | | | 23 (34) | | | 40 (42) | | |
| CC | | | 4 (6) | | | 1 (1) | | |
| ABCC2 | -24 C>T (rs717620) | 5' UTR | | 0.18 | 0.34 | 94 | 0.17 | 0.21 |
| CC | | | 44 (66) | | | 63 (67) | | |
| CT | | | 22 (33) | | | 30 (32) | | |
| TT | | | 1 (1) | | | 1 (1) | | |
| ABCC2 | 3972C>T (rs3740066) | Synonymous, Exon 28 | | 0.34 | 0.81 | 104 | 0.36 | 0.84 |
| CC | | | 30 (45) | | | 43 (41) | | |
| CT | | | 29 (43) | | | 47 (45) | | |
| TT | | | 8 (12) | | | 14 (14) | | |
| SLCO1B1*1b | 388 A>G (rs2306283) | Nonsynonymous, Exon 5 (N130D) | | 0.46 | 0.58 | 84 | 0.40 | 0.73 |
| AA | | | 21 (31) | | | 29 (35) | | |
| AG | | | 31 (46) | | | 42 (50) | | |
| GG | | | 15 (22) | | | 13 (15) | | |
| SLCO1B1*5 | 521 T>C (rs4149056) | Nonsynonymous, Exon 6 (V174A) | | 0.19 | 0.79 | 83 | 0.13 | 0.74 |
| TT | | | 44 (66) | | | 63 (76) | | |
| TC | | | 21 (31) | | | 19 (23) | | |
| CC | | | 2 (3) | | | 1 (1) | | |
| UGT1A1*93 | -3156 G>A (rs10929302) | Intronic | | 0.32 | 0.24 | 103 | 0.28 | 0.57 |
| GG | | | 33 (49) | | | 52 (50) | | |
| GA | | | 25 (37) | | | 44 (43) | | |
| AA | | | 9 (13) | | | 7 (7) | | |
| UGT1A1*28 | TA6>TA7 (rs8175347) | TA repeat, Promoter | | 0.34 | 0.25 | 75 | 0.33 | 0.11 |
| *1/*1 | | | 31 (46) | | | 31 (41) | | |
| *1/*28 | | | 26 (39) | | | 39 (52) | | |
| *28/*28 | | | 10 (15) | | | 5 (7) | | |