IONIC DNA System

Personalized Pharmacogenomic Analysis Report

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■ Variant-Based Drug Risk Summary

rsID	Gene	Drug(s)	Phenot Categ	Signific	Chrom	Positio	Ref	Alt	Genoty	Notes	Summary
rs1801133	MTHFR	atorvastatin	Metabolism/PK	yes	1	11856378	G	A	0/1	NaN	: Metab Clinica signific yes.

rs1801133	MTHFR	l-methylfolate	Efficacy	not stated	1	11856378	G	A	0/1	Please note that alleles have been complemented to the positive strand. Case study of a patient with the AG genotype whose response to pharmacotherap was improved by the addition of folate supplementation	: Efficac Clinica signific not stated

rs1801133	MTHFR	bevacizumab, capecitabine, cisplatin, docetaxel, epirubicin, oxaliplatin, trastuzumab	Efficacy	yes	1	11856378	G	A	0/1	Patients with the AA genotype had decreased progression-free survival and overall survival in multivariable analysis. However, note that these were only a "nominally" significant associations: formally significant was defined as p<0.0026, and nominally significant as p<0.05. No association with response was found. Please note that alleles have been complemented to the plus chromosomal strand.	:: Efficac Clinica signific yes.: Efficac Clinica signific yes.

rs1801133	MTHFR	disulfiram	Efficacy	yes	1	11856378	G	A	0/1	The drop in percentage of cocaine-positive urines for patients with the CT and TT genotypes over the 10 weeks of disulfiram treatment was significantly greater compared to those with the CC genotype. Please note alleles have been complemented to the positive chromosomal strand.	Synop Varian rs1801 in gene MTHF affects respon to disulfir Pheno Efficac Clinica signific yes.

rs1801133	MTHFR	methotrexate	Efficacy	no	1	11856378	G	A	0/1	Alleles were given as C and T. Response measured as relapse-free survival and overall survival.	: Efficac Clinica signific no.
rs1801133	MTHFR	capecitabine	Metabolism/PK	yes	1	11856378	G	A	0/1	as measured by increase in elimination half-life of capecitabine.	:: Metab Clinica signific yes.

rs1801133	MTHFR	benazepril	Efficacy	yes	1	11856378	G	A	0/1	When in a haplotype with rs1801131 allele G, and where only one copy of this GG haplotype was present. Patients with this haplotype had a significantly lower diastolic and systolic blood pressure (DBP and SBP) response to benazepril between baseline and 15 days of treatment, as compared to those with any other haplotype. Please note alleles have been complemented to the plus chromosomal strand.	Synop Varian rs1801 in gene MTHF affects respor to benaz Pheno Efficac Clinica signific yes.

rs1801133	MTHFR	methotrexate	Efficacy	yes	1	11856378	G	A	0/1	Alleles given as C and T. Efficacy of treatment was measured as change of DAS28 from start of treatment to 6months after initiation of treatment. Effect seen in patients receiving monotherapy for methotrexate. In combination with MTHFR A1298C, patients with 677CC-1298CC or 677TT-1298AA had better response to methotrexate by DAS28 change. (p=0.013).	Synop Varian rs1801 in gene MTHF affects respor to metho Pheno Efficac Clinica signific yes.

rs1801133	MTHFR	antipsychotics, chlorpromazine, clozapine, haloperidol, olanzapine, quetiapine, risperidone	Efficacy	yes	1	11856378	G	A	0/1	response is measured by PANSS (Positive and Negative Syndrome Scale), alleles complemented to plus chromosomal strand. In initial evaluation of whole cohort (men and women) authors stated "A; significantly higher mean score was also found in those having MTHFR CT genotype compared with CC and TT; patients. " "Higher negative Positive and Negative Syndrome Scale scores were significantly associated with women and having the CT genotype for MTHFR c.677C>T (β = 4.25; p = 0.008) compared with CC patients. "	: Efficac Clinica signific yes.

rs1801133	MTHFR	methotrexate	Metabolism/PK		1	11856378	G	A	0/1	NaN	:: Metab Clinica signific no.
rs1801133	MTHFR	methotrexate	Metabolism/PK		1	11856378	G	A	0/1	No significant association between this variant and methotrexate concentrations at 72 or 96 hours. Variant described as C677T in the paper and mapped to rs1801133 by PharmGKB. Please note that alleles have been complemented to the positive strand.	:: Metab Clinica signific no.

rs1801133	MTHFR	methotrexate	Efficacy	no	1	11856378	G	A	0/1	No significant association between variant and relapse-free survival, overall survival or event-free survival. Please note that alleles have been complemented to the positive strand.	:	Efficac Clinica signific no.

rs1801133	MTHFR	methotrexate	Metabolism/PK	yes	1	11856378	G	A	0/1	Patients carrying at least one A allele had significantly higher AUC of methotrexate than patients with the GG genotype. Please note that alleles have been complemented to the positive strand.	:: Metab Clinica signific yes.
rs1801133	MTHFR	methotrexate	Metabolism/PK	yes	1	11856378	G	A	0/1	Please note that alleles have been complemented to the positive strand.	:: Metab Clinica signific yes.

rs1801133	SLCN6, MTHFR	methotrexate	folate metabolism	mPK	1	11856378	G	A	0/1	Please note that alleles have been complemented to the positive strand.	:

rs1801133	MTHFR	methotrexate	Efficacy	yes	1	11856378	G	A	0/1	Patients with the AA genotype had better overall survival than patients with the AG or GG genotypes. Variant referred to in the paper as C677T. Please note that alleles have been complemented to the positive strand.	Synop Varian rs1801 in gene MTHF affects respor to metho Pheno Efficac Clinica signific yes.

rs1801133	MTHFR	methotrexate	Efficacy	no	1	11856378	G	A	0/1	No significant difference in relapse-free survival between genotype groups. Please note that alleles have been complemented to the positive strand.	: Clinica no.	Efficac signific

rs1801133	SLCN6, MTHFR	methotrexate	Metabolism/PK	1	11856378	G	A	0/1	Please note that alleles have been complemented to the positive strand. This variant is referred to as 677C>T is the paper and was mapped to rs1801133 by PharmGKB. The AA genotype was not found in the cohort.	:	

rs1801133	SLCN6, MTHFR	methotrexate	Metabolism/PK	no	1	11856378	G	A	0/1	Please note that alleles have been complemented to the positive strand.	:
rs1801133	MTHFR	methotrexate	Metabolism/PK	yes	1	11856378	G	A	0/1	SNP is referred to in the paper as 677 C>T and was mapped to rs1801133 by PharmGKB. Please note that alleles have been complemented to the positive strand.	:: Metab Clinica signifi yes.

rs1801133	MTHFR	methotrexate	Metabolism/PK	yes	1	11856378	G	A	0/1	Please note that alleles have been complemented to the positive strand. Patients with the AG genotype did not have significantly different methotrexate plasma levels compared to those with the GG genotype.	:: Metab Clinica signific yes.

rs1801133	MTHFR	methotrexate	Metabolism	PD/PK	1	11856378	G	A	0/1	No significant difference in allele frequency between patients with delayed methotrexate excretion and those with normal methotrexate excretion.	:: Metab Clinica signific no.

rs1801133	MTHFR	methotrexate	Efficacy	no	1	11856378	G	A	0/1	alleles complemented to plus chromosomal strand. Response measured by ESR, erythrocyte sedimentation rate; TJC, tender joints counts; SJC, swollen joints counts; DAS28, Disease Activity Score in 28 joints. "DAS28 was decreased after the post-treatment in 677TT and 1298AC, but was not statistically significant."	:	Effica Clinica signific nal

rs1801133	MTHFR	capecitabine, fluorouracil	Dosage	yes	1	11856378	G	A	0/1	"differences were not present in male patients."	Dosag Clinica signific yes.
rs1801133	MTHFR	methotrexate	Toxicity, Metabolism/PK	yes	1	11856378	G	A	0/1	Half life and AUC was significantly increased and elimination rate, volume of distribution and total body clearance was significantly decreased.	:: Toxicit Metab Clinica signific yes.
rs1801133	MTHFR	capecitabine, fluorouracil, leucovorin, oxaliplatin	Efficacy	yes	1	11856378	G	A	0/1	FOLFOX : (54.4%) and CAPOX (45.6%) regimens. Measured as cumulative survival over 100 months since diagnosis.	Efficac Clinica signific yes.

rs1801133	MTHFR	risperidone	Efficacy	no	1	11856378	G	A	0/1	Please note that alleles have been complemented to the positive strand.	: Efficac Clinica signific no.
rs1801133	MTHFR	methotrexate	Efficacy	no	1	11856378	G	A	0/1	Please note: alleles have been complemented to the plus chromosomal strand.	: Efficac Clinica signific no.
rs1801133	MTHFR	methotrexate	Efficacy	no	1	11856378	G	A	0/1	Please note: alleles have been complemented to the plus chromosomal strand.	: Efficac Clinica signific no.
rs1801133	MTHFR	mercaptopurine methotrexate	Dosage	no	1	11856378	G	A	0/1	NaN	:: Dosag Clinica signific no.

rs1801133	MTHFR	methotrexate	Toxicity	no	1	11856378	G	A	0/1	Discontinuation due to adverse effects. Please note that alleles have been complemented to the plus chromosomal strand.	1	rs1801 in gene MTHF affects respon to metho Clinica signific noted

rs1801133	MTHFR	antineoplastic agents	Efficacy	no	1	11856378	G	A	0/1	No significant difference in response rate or progression-free survival time was seen between the genotypes. Patients were either receiving FOLFOX/XELOX or FOLFIRI regimens (respectively: fluorouracil, leucovorin, oxaliplatin; capecitabine, oxaliplatin; fluorouracil, leucovorin, irinotecan). Please note alleles have been complemented to the plus chromosomal strand.	: Efficac Clinica signific no.

rs1801133	MTHFR	methotrexate	Efficacy	no	1	11856378	G	A	0/1	NaN	: Efficac Clinica signific no.
rs1801133	MTHFR	methotrexate	Efficacy	yes	1	11856378	G	A	0/1	"MTHFR 677TT ... associated with over 4-fold increased risk for nonresponse " Alleles complement to plus chromosomal strand.	Synop Varian rs1801 in gene MTHF affects respor to metho Phenc Efficac Clinica signific yes.
rs1801133	MTHFR	methotrexate	Metabolism/PK	yes	1	11856378	G	A	0/1	AA vs GG and AG vs GG were statistically significant.	:: Metab Clinica signific yes.

rs1801133	MTHFR	Platinum compounds	Efficacy	no	1	11856378	G	A	0/1	Patients were treated with cisplatin or carboplatin in combination with a third-generation drug (gemcitabine, paclitaxel, pemetrexed or vinorelbine). No significant association with overall response rate (ORR), overall survival (OS) or progression-free survival (PFS) was found for this SNP. Please note that alleles have been complemented to the plus chromosomal strand.	: Efficac Clinica signific no.

rs1801133	MTHFR	folic acid, hydroxychloroquine, methotrexate, sulfasalazine	Efficacy	no	1	11856378	G	A	0/1	NaN	: Efficac Clinica signific no.
rs1801133	MTHFR	methotrexate	Efficacy	no	1	11856378	G	A	0/1	NaN	: Efficac Clinica signific no.
rs1801133	MTHFR	methotrexate	Efficacy	no	1	11856378	G	A	0/1	In this study those with a "favourable" MTHFR genotype (ie. 677CC/ rs1801133 or 1298AA/rs1801 were treated with a higher dose.	: Efficac Clinica signific no.

rs1801133	MTHFR	folic acid	Metabolism/PK	not stated	1	11856378	G	A	0/1	Please note that alleles have been complemented to the positive stand.: Case study of a patient with the A allele at rs1801133 and the G allele at rs1801131 who subsequently responded to folate supplementation	:: rs1801 in gene MTHF affects respo to folic acid. Pheno Metab Clinica signific not stated

rs1801133	MTHFR	benazepril	Efficacy	no	1	11856378	G	A	0/1	No significant differences in the change of systolic blood pressure between baseline and after 15 days of benazepril treatment was seen between the genotypes. Please note alleles have been complemented to the plus chromosomal strand.	Synop Varian rs1801 in gene MTHF affects respor to benaz Pheno Efficac Clinica signific no.

rs1801133	MTHFR	benazepril	Efficacy	yes	1	11856378	G	A	0/1	Patients with the AA genotype had a greater change in diastolic blood pressure between baseline and after 15 days of treatment, compared to those with the AG and GG genotypes. Please note alleles have been complemented to the plus chromosomal strand.	Synop Varian rs1801 in gene MTHF affects respor to benaz Pheno Efficac Clinica signific yes.

rs1801133	MTHFR	methotrexate	Efficacy	no	1	11856378	G	A	0/1	Also, with increasing copies of rs1801133 G, there was also an increase in the association between rs1801131 and methotrexate response.	: Efficac Clinica signific no.
rs1801133	CLCN6, MTHFR	methotrexate	Efficacy	yes	1	11856378	G	A	0/1	Non-response was classified by DAS28 >3.2 at two consecutive evaluations (minimum period of at least 6 months of MTX therapy).	Syste Varian rs1801 in gene CLCN MTHF affects respon to metho Pheno Efficac Clinica signific yes.

rs1801133	MTHFR	methotrexate	Efficacy	yes	1	11856378	G	A	0/1	Authors say this variant is associated with non-response in recessive model (minor allele/minor allele vs minor/major + major major and describe variant as C677T). Alleles complemented to plus chromosomal strand.	Synop Varian rs1801 in gene MTHF affects respor to metho Pheno Efficac Clinica signific yes.
rs1801133	MTHFR	fluorouracil, leucovorin, oxaliplatin	Efficacy	yes	1	11856378	G	A	0/1	NaN	: Efficac Clinica signific yes.
rs1801133	MTHFR	methotrexate	Efficacy	no	1	11856378	G	A	0/1	NaN	: Efficac Clinica signific no.

rs1801133	MTHFR	methotrexate	Efficacy	no	1	11856378	G	A	0/1	NaN	: Efficac Clinica signific no.
rs1801133	MTHFR	methotrexate	Toxicity, Metabolism/PK	no	1	11856378	G	A	0/1	NaN	:: Toxicit Metab Clinica signific no.
rs1801133	MTHFR	methotrexate	Metabolism/PK	yes	1	11856378	G	A	0/1	NaN	:: Metab Clinica signific yes.

rs1801133	MTHFR	methotrexate	Efficacy	no	1	11856378	G	A	0/1	Genotype: AA is associated with increased red blood cell folate when treated with methotrexate as compared to genotype GG but this did not correlate with disease activity or red blood cell methotrexate metabolites.	Efficac Clinica signific no.

rs1801133	MTHFR	methotrexate	Toxicity	yes	1	11856378	G	A	0/1	Meta-analysis with 8 studies. Cohorts of pediatrics (6 studies) and adults (2 studies) were also considered separately. In the pediatric and overall cohort, those with the AA genotype had a greater risk of experiencing relapse as compared to those with the AG or GG genotype; no significant result was seen when considering the adult cohort (the authors note that more studies are needed before reliable conclusions can be drawn regarding the influence of rs1801133 on relapse in adults with ALL). Please note alleles have been complemented to the plus chromosomal strand.	:::::: Varian rs1801 in gene MTHF affects respor to metho Clinica signific yes.

rs1801133	MTHFR	Vitamin B-complex, Incl. Combinations	Efficacy	yes	1	11856378	G	A	0/1	Please note that allele has been complemented to the positive strand. Patients who carried either the A allele of rs1801133 or the G allele of rs1801131 were recruited into the study (genotypes are not given) and given a capsule containing reduced B vitamins and macronutrients or a placebo. Patients taking the B vitamin capsule showed a significant decrease in MADRS score over 8 weeks of treatment compared to those taking placebo.	: Varian rs1801 in gene MTHF affects respor to Vitami B-com Incl. Combi Pheno Efficac Clinica signifi yes.

rs1801133	MTHFR	fluorouracil	Efficacy	yes	1	11856378	G	A	0/1	Patients receiving fluorouracil chemotherapy. Response included complete response (CR) and partial response (PR). Non-response included progressive disease (PD) and stable disease (SD). Please note that alleles have been complemented to the plus chromosomal strand.	::s:: Variant is a gene affects respon to fluorou Pheno Efficac Clinica signific yes.

rs1801133	MTHFR	methotrexate	Metabolism/PK	yes	1	11856378	G	A	0/1	Alleles complemented "As depicted in Fig. 1, the MTHFR 677CT/TT genotype exhibited a higher likelihood of delayed clearance compared with the wild-type (OR = 3.056/3.456, P = 0.019/0.009)."	::	Metab Clinica signific yes.
rs1801133	CLCN6, MTHFR	tenofovir	Metabolism/PK	no	1	11856378	G	A	0/1	Significance threshold was set at 4.5E-3.	:	Metab Clinica signific no.
rs1801133	MTHFR	vitamin b12 and folic acid	Efficacy	yes	1	11856378	G	A	0/1	Full text was unavailable; annotation made using abstract.	:	Efficac Clinica signific yes.

rs1801133	MTHFR	methotrexate	Efficacy	yes	1	11856378	G	A	0/1	"In the multivariable analysis, which included smoking, the DHFR rs408626 and MTHFR rs1801133 variants retained a statistically significant association with MTX failure (OR 3.12, P = .017 and OR 2.86, P = .015, respectively in a dominant model). The DHFR rs408626-G and MTHFR rs1801133-C alleles were associated with a higher risk of MTX failure. "	Synop Varian rs1801 in gene MTHF affects respor to metho Pheno Efficac Clinica signific yes. Alleles complemented.

rs2231142	ABCG2	rosuvastatin	Efficacy	no	3	141313677	G	T	0/0	ABCG2 421C>A did not significantly affect the lipid-lowering response of rosuvastatin.	: Efficac Clinica signific by.
rs2231142	ABCG2	allopurinol	Efficacy	yes	3	141313677	G	T	0/0	this was significant in both LASSO and New Zealand cohorts and meta-analysis. Good responders were defined as SU <6 mg/dl on allopurinol ■300 mg/day and poor responders as SU ■6 mg/dl despite allopurinol >300 mg/day	Synop Varian rs2231 in gene ABCG affects respon to allopu Pheno Efficac Clinica signific yes.

rs2231142	ABCG2	rosuvastatin	Metabolism/PK	not stated	3	141313677	G	T	0/0	NaN	: Metab Clinica signific not stated
rs2231142	ABCG2	rosuvastatin	Metabolism/PK	yes	3	141313677	G	T	0/0	NaN	:: Metab Clinica signific yes.

rs2231142	ABCG2	rosuvastatin	Metabolism/PK	yes	3	141313677	G	T	0/0	"The mean Css/D of RST and its metabolites were significantly higher in the subjects carrying the ABCG2 421A than in non-carriers of this; allele. The effects of this allele remained significant after being adjusted by the baseline characteristics and false discovery rate; (FDR) (Padj < 0.01, FDR < 0.05)."	:: Metab Clinica signific yes.

rs2231142	ABCG2	rosuvastatin	Metabolism/PK	yes	3	141313677	G	T	0/0	This variant affected rosuvastatin concentrations significantly and potentially affect serum levels of pro-inflammatory and pro-angiogenic markers.	:: Metab Clinica signific yes.
rs2231142	ABCG2	apixaban	Metabolism/PK	yes	3	141313677	G	T	0/0	was significant for 3 measures of PK : AUCss, Cmax,ss, and Cmin,ss.	:: Metab Clinica signific yes.
rs2231142	ABCG2	pitavastatin	Metabolism	no	PK	3	141313677	G	T	0/0	Please note that alleles have been complemented to the positive strand.	:

rs2231142	ABCG2	Opioid anesthetics, Other general anesthetics, volatile anesthetics	Efficacy	yes	3	141313677	G	T	0/0	The TT genotype was significantly associated with a shorter recovery time from general anesthesia compared to the GG and GT genotypes.	:: rs2231 in gene ABCG effects respon to Opioid anesth Other genera anesth volatile anesth Pheno Efficac Clinica signifi

rs2231142	ABCG2	gefitinib	Metabolism/PK	3		141313677	G	T	0/0	No significant difference in area under the plasma concentration-tir curve (AUC) or maximum plasma concentration (Cmax) or trough concentrations (C0) was seen between the genotypes. Please note that alleles have been complemented to the plus chromosomal strand.	:

rs2231142	ABCG2	sulfasalazine	Metabolism/PK	yes	3	141313677	G	T	0/0	A single oral dose was given and pharmacokinetic parameters were measured at different times points. Parameters AUC0-48, Cmax and CLtotal/F were all significantly different in individuals with the GT genotype compared to GG, and in individuals with the TT genotype compared to GG and compared to GT. Mean plasma concentrations of Sulfasalazine at all time points were significantly higher in TT subjects compared to GG (0-48 hours, except 0.5 hours).	:: Metab Clinica signific yes.

rs2231142	ABCG2	gefitinib	Metabolism/PK		3	141313677	G	T	0/0	No significant difference in median area under the concentration-tir curve from 0 to 24 hours (AUC0-24; p=0.323) or trough plasma concentration (C0; p=0.429) was seen between the two genotype groups. Please note that alleles have been complemented to the plus chromosomal strand.	:

rs2231142	ABCG2	N-desmethyltadalafil	Metabolism/PK	3	141313677	G	T	0/0	Alleles complemented to plus chromosomal strand. RS number has typo in paper (effect is reported for ABCG2 c.421C>A and lists in methods as rs2231141.	:

rs2231142	ABCG2	sunitinib	Efficacy	no	3	141313677	G	T	0/0	The genotype was not associated with progression-free survival, or overall survival, or clinical benefit. Clinical benefit was defined as either partial response or stable disease. Please note, alleles have been complemented to the + chromosomal strand.	Synop Varian rs2231 in gene ABCG affects respor to sunitin

rs2231142	ABCG2	lamotrigine	Metabolism/PK		3	141313677	G	T	0/0	Please note that alleles have been complemented to the positive strand.	:
rs2231142	ABCG2	lamotrigine	Dosage	no	3	141313677	G	T	0/0	Please note that alleles have been complemented to the positive strand.	: Dosag Clinica signific no.

rs2231142	ABCG2	methotrexate	Metabolism/PK		3	141313677	G	T	0/0	No significant association between this variant and methotrexate concentrations at 72 or 96 hours. Variant described as C421A in the paper and mapped to rs2231142 by PharmGKB. Please note that alleles have been complemented to the positive strand.	:

rs2231142	ABCG2	rosuvastatin	Efficacy	yes	3	141313677	G	T	0/0	Volunteers with ABCG2 c.421C>A genotype exhibited significantly higher mean % changes in LDL, apoB and total cholesterol levels as compared to those with the wild-type genotype.	Efficac Clinica signific yes.
rs2231142	ABCG2	methotrexate	Metabolism/PK		3	141313677	G	T	0/0	NaN	:

rs2231142	ABCG2	leflunomide	Efficacy	no	3	141313677	G	T	0/0	"79 patients (74 females and five males), ... Seventy patients (88.6% of patients) were receiving leflunomide only; five patients (6.3%) were receiving additionally prednisolone, while nine patients (11.4%) were receiving combined hydroxychloroquine and leflunomide." Authors compared genotypes between "Controlled" RA, DAS 28 score of 3.2 or less and "Poorly controlled" RA with DAS 28 of 3.2 and above.	Synop Varian rs2231 in gene ABCG affects respor to leflunc

rs2231142	ABCG2	methotrexate	Metabolism	PK	3	141313677	G	T	0/0	No significant difference in allele frequency between patients with delayed methotrexate excretion and those with normal methotrexate excretion.	:

rs2231142	ABCG2	methotrexate	Metabolism/PK	yes	3	141313677	G	T	0/0	Please note that alleles have been complemented to the positive strand. Patients with the TT genotype did not have significantly different methotrexate plasma levels compared to those with the GG genotype.	: rs2231 in gene ABCG affects respo to metho Pheno Metab Clinica signific yes.
rs2231142	ABCG2	pazopanib	Metabolism/PK	no	3	141313677	G	T	0/0	Alleles complemented to plus chromosomal strand.	:: Metab Clinica signific no

rs2231142	ABCG2	rivaroxaban	Metabolism/PK		3	141313677	G	T	0/0	Alleles complemented "The majority of patients had taken rivaroxaban 15 mg" Patients had "NVAF undergoing AF catheter ablation".	:: Metab Clinica signific no.
rs2231142	ABCG2	pitavastatin	Metabolism/PK		3	141313677	G	T	0/0	ABCG2 421C4A variant did not appear to be associated with the altered pharmacokinetic of pitavastatin.	:

rs2231142	ABCG2	imatinib	Metabolism/PK	yes	3	141313677	G	T	0/0	"The C0/D of IM appeared higher in patients with the rs2231142 mutant T-allele (3.98 ± 1.36 ng/ml·mg-1) than that in wild-type GG genotype patients (3.40 ± 1.21 ng/ml·mg-1)."	: rs2231 in gene ABCG affects respor to imatini Pheno Metab Clinica signific yes.
rs2231142	ABCG2	rosuvastatin	Metabolism/PK	yes	3	141313677	G	T	0/0	Alleles complement to plus chromosomal strand. Meta-analysis of 8 studies looking at AUC and Cmax of rosuvastatin.	:: Metab Clinica signific yes.

rs2231142	ABCG2	dolutegravir	Metabolism/PK	yes	3	141313677	G	T	0/0	Alleles complemented to plus chromosomal strand. "ABCG2 c.421C>A (rs2231142) was independently associated with a 28% increase in dolutegravir Cmax; (b = 0.053; P = 0.047) in the homozygous variant. GM Cmax (95% CI); was 3893 (3774–4240) 4346 (3629–5531) and 4994 (single value) ng/mL in the CC, CA and AA genotype groups, respectively." "Pooled samples from three Phase I clinical trials (NCT02219217,; NCT02509195 and NCT03094507) and one Phase III clinical trial; (NCT02351908) carried out at the St Stephen's AIDS Trust clinical trial unit,; London" From Table one, only one individual was homozygous for minor allele.	: a recom

rs2231142	ABCG2	felodipine	Metabolism/PK	yes/	3	141313677	G	T	0/0	NaN	: rs2231 in gene ABCG affects respon to felodip Pheno Metab Clinica signifi yes.
rs2231142	ABCG2	apixaban	Metabolism/PK	yes/	3	141313677	G	T	0/0	GG genotype has higher concentration ratio of apixaban.	:: Metab Clinica signifi yes/do

rs2231142	ABCG2	allopurinol	Efficacy	yes	3	141313677	G	T	0/0	The authors designated "response to allopurinol" as a reduction of serum uric acid levels to below 6 mg/dL. Mean age was 68 years, 75% were male. The elevated mean baseline SUA before treatment was 8.9 mg/dL. No other SNPs reached genome wide significance.	Synop Varian rs2231 in gene "ABCG affects respor to allopu Pheno Efficac Clinica signific yes.

rs2231142	ABCG2	dolutegravir	Metabolism/PK	yes	3	141313677	G	T	0/0	mean peak plasma concentration	: a recom

rs2231142	ABCG2	axitinib	Metabolism/PK	yes	3	141313677	G	T	0/0	The authors develop a prediction model and calculated area under the concentration curve (AUC) using 6 SNPs (rs17868323, rs3832043, rs2231142, rs2032582, rs1045642, rs35305980) was compared with actual AUC in 16 patients prospectively which significantly correlated with the objective response rate (P = 0.0002), hand-foot syndrome, P = 0.0055 and hypothyroidism, P = 0.0381, and correlated with actual AUC (P < 0.0001) - the validation study, calculated AUC prior to axitinib treatment precisely predicted actual AUC after axitinib treatment (P = 0.0066).	:: Metab Clinica signific yes.

rs2231142	ABCG2	tacrolimus	Dosage	no	3	141313677	G	T	0/0	No significant difference in daily dose of tacrolimus was seen between genotypes of the rs2231142 SNP, at any point between 0 and 60 days after transplant. Please note alleles have been complemented to the plus chromosomal strand.	: Dosag Clinica signifi no.

rs2231142	ABCG2	imatinib	Dosage	no	3	141313677	G	T	0/0	No significant association between genotype and chance of requiring an imatinib dose reduction. Please note that alleles have been complemented to the positive strand.

rs2231142	ABCG2	allopurinol	Efficacy	yes	3	141313677	G	T	0/0	Good response was defined as serum urate of < 6 mg/dl on a dose of allopurinol on less than 300 mg/day and poor response was defined as serum urate of greater than or equal to 6 mg/dl despite allopurinol of greater than 300 mg/d. The frequency of the GT and TT genotypes was higher in the poor responder group (N=68) than in the good responder group (N=120). This association remained significant even after adjusting for gender, BMI, ethnicity, estimated glomerular filtration rate, diuretic use, and serum urate concentration at baseline.	Synop Varian rs2231 in gene ABCG affects respor to allopu Pheno Efficac Clinica signifi yes.

rs2231142	ABCG2	tacrolimus	Dosage	no	3	141313677	G	T	0/0	No significant difference in clearance or blood concentration of tacrolimus was seen between the genotypes of the rs2231142 SNP, at any point between 0 and 60 days after transplant. Please note alleles have been complemented to the plus chromosomal strand.	: Dosag Clinica signifi no.

rs2231142	ABCG2	fluorouracil, irinotecan, oxaliplatin	Efficacy	no	3	141313677	G	T	0/0	Patients were taking either IFL (irinotecan + fluorouracil + leucovorin; n=114), FOLFOX (fluorouracil + oxaliplatin + leucovorin; n=299) or IROX (irinotecan + oxaliplatin; n=107). No significant association was seen between this variant and confirmed response rate, overall survival or time to progression in any of the treatment groups OR all treatment groups considered together. Significance level was set at 0.01.	: Efficac Clinica signific no.

rs2231142	ABCG2	methotrexate	Toxicity	no	3	141313677	G	T	0/0	Discontinuation due to adverse effects. Please note that alleles have been complemented to the plus chromosomal strand.	rs2231 in gene ABCG affects respon to metho Clinica signific not

rs2231142	ABCG2	sulfasalazine	Efficacy	yes	3	141313677	G	T	0/0	After adjusting for age, baseline DAS28, smoking status and shared epitope positivity, those who carried one or more T allele were more likely to achieve remission after 12 months of treatment. Remission defined as an SDAI of <3.3. Please note that alleles have been complemented to the positive chromosomal strand.	: Efficac Clinica signific yes.

rs2231142	ABCG2	antineoplastic agents	Efficacy	yes	3	141313677	G	T	0/0	Patients with the GG genotype receiving a FOLFOX/XELOX regimen (respectively: fluorouracil, leucovorin, oxaliplatin, capecitabine, oxaliplatin) had a decreased response rate, as compared to those with the GT or TT genotype. No significant difference in response rate was seen in patients receiving a FOLFIRI regimen (fluorouracil, leucovorin, irinotecan). No significant differences in progression-free survival were seen for either regimen. Please note alleles have been complemented to the plus chromosomal strand.	: a synthe of the gene ABCG and the role of the gene ABCG in the respor to antine agents

rs2231142	ABCG2	granisetron, palonosetron	Efficacy	no	3	141313677	G	T	0/0	There was no significant association between this variant and proportion of patients showing a "complete response" to antiemetic treatment during the first 120 hours after cisplatin-based chemotherapy initiation. Complete response defined as no vomiting episodes or no use of rescue medication. Patients were either receiving granisetron (n=79) or palonosetron (n=77), both in combination with dexamethasone and aprepitant. Please note that alleles have been complemented to the plus chromosomal strand.	: Efficac Clinica signific no.

rs2231142	ABCG2	lamotrigine	Efficacy	no	3	141313677	G	T	0/0	Alleles given as A and C. Efficacy was determined by monitoring the frequency of patients' epileptic seizures within one year. Each patient was evaluated at 4 weeks after treatment initiation and then at 3-month intervals thereafter. The difference in seizure frequency was based on the difference between the 3-month retrospective baseline frequency and the seizure frequency at 12-month visit, which was reported for the last 3 months prior to the last visit. Good efficacy was defined as seizure-free or a 50% or greater reduction in seizure frequency within a 1-year follow-up period.	Synop Varian rs2231 in gene ABCG affects respor to lamotr Pheno Efficac Clinica signific no.

rs2231142	ABCG2	lamotrigine	Metabolism/PK	yes	3	141313677	G	T	0/0	Lamotrigine concentrations were measured as steady state in plasma in the early morning before breakfast after at least one month of continuous treatment with lamotrigine monotherapy. Alleles given as A and C.	Lamotrigine concentrations

rs2231142	ABCG2	sunitinib	Metabolism/PK	yes	3	141313677	G	T	0/0	The dose-normalized area under the plasma concentration-time curve from 0 to 24 hours (AUC) for the composite of sunitinib + SU12662 (its active metabolite) was highest for those with the TT genotype, followed by those with the GT genotype then those with the GG genotype (TT>GT>GG). This SNP was identified as a statistically significant variability factor for composite exposure. Please note that alleles have been complemented to the plus chromosomal strand.	: rs2231 in gene ABCG affects respor to sunitin Pheno Metab Clinica signifi yes.

rs2231142	ABCG2	lamotrigine	Metabolism/PK	yes	3	141313677	G	T	0/0	Alleles given as A and C. The concentration was measured as lamotrigine trough concentration / dose normalized by body weight. Included patients had been on lamotrigine monotherapy for at least a month with complete medical records, had normal renal and hepatic functions, and had therapeutic drug monitoring with good compliance.	: a recom

rs2231142	ABCG2	lamotrigine	Metabolism/PK		3	141313677	G	T	0/0	NaN	:
rs2231142	ABCG2	deferasirox	Metabolism/PK		3	141313677	G	T	0/0	The genotype was not associated with area under the concentration curve (AUC) above the effectiveness cutoff of 360 micrograms/mL as compared in univariate analysis.	Synop Varian rs2231 in gene ABCG affects respon to defera Pheno Metab Clinica signifi ass
rs2231142	ABCG2	acetaminophen	Metabolism/PK		3	141313677	G	T	0/0	There were no TT homozygotes and only 7 heterozygotes.	: Metab Clinica signifi no

rs2231142	ABCG2	hmg coa reductase inhibitors	Efficacy	yes	3	141313677	G	T	0/0	as part of a three SNP genetic risk score with rs10455872 in LPA and rs2075650 in APOE. Associated allele not explicitly stated but methods reference Tomlinson et al. so used the associated allele from there [PMID:20130565	: Efficac Clinica signific yes.
rs2231142	ABCG2	risperidone	Metabolism/PK	yes	3	141313677	G	T	0/0	Alleles complemented to plus chromosomal strand.	: rs2231 in gene ABCG affects respon to risperi

rs2231142	ABCG2	allopurinol	Dosage	yes	3	141313677	G	T	0/0	ABCG2 genotype and diuretic use explained 53% of the variability in prediction error.	: Dosag Clinica signific yes.

rs2231142	ABCG2	tenofovir	Metabolism/PK	yes	3	141313677	G	T	0/0	Patients with the GT or TT genotype had a 1.51-fold increase in tenofovir area under the concentration-time curve (AUC) as compared to those with the GG genotype. Multivariable model controlling for age (per decade), body mass index (per 10 percent increase), African American race, ritonavir use, and whether eGFR is less than 70 ml/min per 1.73m2	: Metab Clinica signific yes.

rs2231142	ABCG2	antiepileptic	Efficacy	no	3	141313677	G	T	0/0	Please note, alleles have been complemented for the plus strand. Epilepsy patients on antiepileptic drug (AED) treatment were divided into those with drug-responsive or drug-resistant epilepsy and genotypes and allele frequencies were compared. No significant association found with this variant.	: Efficac Clinica signific no.

rs2231142	ABCG2	sulfasalazine	Metabolism/PK	not stated	3	141313677	G	T	0/0	The mean AUC(0,48 h) was 3-fold higher and Cmax was 2.5-fold higher in individuals with the TT genotype as compared to individuals with the GG genotype. The apparent oral clearance (dose/ AUC(0,48 h)) was 3-fold lower in individuals with the TT genotype as compared to individuals with the GG genotype. The authors reported no differences in sulfasalazine PK between the GG genotype and GT genotype, however a single individual was compound heterozygous at rs72552713 (CT) and rs2231142 (GT) in and altered PK as compared to other individuals who were heterozygous only at rs2231142 (GT). Please note: alleles have been complemented to the + chromosomal strand.	: Metab Clinica signific not stated

rs2231142	ABCG2	atorvastatin	Efficacy	no	3	141313677	G	T	0/0	The authors tested whether differences in the following biomarkers that corresponded to genotype (dominant model): triglycerides, HDL-cholsterol, LDL-cholesterol and total cholesterol.	: Efficac Clinica signific so.

rs2231142	ABCG2	imatinib	Other, Metabolism/PK	yes	3	141313677	G	T	0/0	The association was with decreased clearance. Genotype alone was not associated, but genotype enhanced a more major association with body weight, albuminemia and plasma alpha1-acid glycoprotein. There were no TT in this small group(41 GG, 5 GT).

rs2231142	ABCG2	atorvastatin	Metabolism/PK		3	141313677	G	T	0/0	The AUC of atorvastatin in patients carrying ABCG2 c.421AA tended to be smaller compared with those in patients with c.421CC or c.421CA after its microdosing and therapeutic dosing. However, it did not reach statistical significance.	: Metab Clinica signific no.

rs2231142	ABCG2	imatinib	Efficacy	yes	3	141313677	G	T	0/0	Direction of relationship not explicitly stated. Assumed minor allele as T and having lower odds ratio of response. Alleles complemented to plus strand as gene is on minus chromosomal strand.	Synop Varian rs2231 in gene ABCG affects respor to imatini Pheno Efficac Clinica signific yes.

rs2231142	ABCG2	lamotrigine	Metabolism/PK	yes	3	141313677	G	T	0/0	In patients with the AA and AG genotype, steady-state measured and dose-adjusted lamotrigine trough concentration was higher in those administered lamotrigine as compared to the GG genotype and lower in those administered lamotrigine only as compared to the GG genotype.	: a recom

rs2231142	ABCG2	selumetinib	Metabolism/PK	no	3	141313677	G	T	0/0	Not associated with normalized dose when allele was assessed within ethnic groups (Asian, White, Black) and when all ethnic groups were pooled together.	:
rs2231142	ABCG2	iguratimod	Efficacy	yes	3	141313677	G	T	0/0	Alleles complementary to plus strand.	: a synthetic of the rs2231 gene ABCG in the gene ABCG .
rs2231142	ABCG2	apixaban	Metabolism/PK	not stated	3	141313677	G	T	0/0	NaN	: Metab Clinica signific not stated

rs2231142	ABCG2	methotrexate	Metabolism/PK	yes	3	141313677	G	T	0/0	The T allele is associated with longer half-life of methotrexate.	: rs2231 in gene ABCG affects respon to metho Pharmo Metab Clinica signific yes.
rs2231142	ABCG2	sulfasalazine	Dosage	no	3	141313677	G	T	0/0	Exposure was measured as Cmax and AUC 0-24.	: Dosag Clinica signific no.
rs2231142	ABCG2	rosuvastatin	Metabolism/PK	no	3	141313677	G	T	0/0	Exposure was measured as Cmax and AUC 0-24.	: Metab Clinica signific no.
rs2231142	ABCG2	sunitinib	Metabolism/PK	no	3	141313677	G	T	0/0	as measured by C/D Ratio or total Sunitinib Dose-Adjusted Concentration.	:

rs2231142	ABCG2	talinolol	Metabolism/PK	no	3	141313677	G	T	0/0	This was a twin study of monozygotic and dizygotic twins.	: Metab Clinica signific no.
rs2231142	ABCG2	gefitinib	Efficacy	no	3	141313677	G	T	0/0	NaN	Synop Varian rs2231 in gene ABCG affects respon to gefitini
rs2231142	ABCG2	rosuvastatin	Efficacy	yes	3	141313677	G	T	0/0	as measured by achievement of their LDL cholesterol target (significance given for variants of either CYP3A5*1, 6986A>G, rs776746 and ABCG2; 421C>A, rs2231142).	: Efficac Clinica signific yes.

rs2231142	ABCG2	granisetron, palonosetron	Efficacy	no	3	141313677	G	T	0/0	Patients with cancer, treated with cisplatin who had been treated with granisetron and palonsetron. Nausea and vomiting episodes were recorded for the first 120 h post cisplatin. With regard to vomiting events, presence/absence of symptoms and the time of onset were described. Nausea severity was categorized using a 4-point Likert scale (0 = no nausea, 1 = mild nausea, 2 = moderate nausea and 3 = severe nausea) according to the subjective assessment of each patient.	: Efficac Clinica signific no.

rs2231142	ABCG2	sunitinib	Metabolism/PK	no	3	141313677	G	T	0/0	NaN	:
rs2231142	ABCG2	sorafenib	Metabolism/PK	yes	3	141313677	G	T	0/0	measured by concentration/(dose weight) ratio.; (alleles complemented to plus strand). No TT homozygotes were observed.	Pheno Metab Clinic/(signifi yes.

rs2231142	ABCG2	n-desethyl sunitinib, sunitinib	Toxicity, Metabolism/PK	no	3	141313677	G	T	0/0	in a single case study. Authors state "Therefore, we speculated that the extremely high plasma concentrations of sunitinib and SU12662 caused by the ABCG2 421 AA genotype might have resulted in severe toxicities to the patient."	Synop Varian rs2231 in gene ABCG affects respon to n-dese sunitin sunitin Pheno Toxicit Metab Clinica signific

rs2231142	ABCG2	ceftriaxone	Metabolism/PK	3	141313677	G	T	0/0	Please note that alleles have been complemented to the positive strand. No significant association was found between this variant and concentrations of ceftriaxone in the plasma or cerebrospinal fluid (CSF) of patients. There was also no significant association between this variant and ceftriaxone CSF to plasma ratios.	:

rs2231142	ABCG2	letermovir	Metabolism	mPK	3	141313677	G	T	0/0	No significant association between this variant and letermovir AUC.	:
rs2231142	ABCG2	selumetinib	Metabolism	mPK	3	141313677	G	T	0/0	Not associated with AUC, AUC0-12 when allele was assessed within ethnic groups (Asian, White, Black) and when all ethnic groups were pooled together. Only P-values for AUC presented here.	:

rs2231142	ABCG2	sulfasalazine	Other	no	3	141313677	G	T	0/0	No significant difference in likelihood of cessation of sulphasalazine treatment due to adverse effects was seen between those who carried the T allele and those who did not. Most common adverse events were nausea/vomiting and fatigue/lethargy; please refer to paper for full list. Note that alleles have been complemented to the plus chromosomal strand.	:: rs2231 in gene ABCG affects respon to sulfasa Pheno Other. Clinica signifi no.

rs2231142	ABCG2	methotrexate	Metabolism/PK		3	141313677	G	T	0/0	TT vs GG and TG vs GG were not statistically significant.	:
rs2231142	ABCG2	voriconazole	Metabolism/PK		3	141313677	G	T	0/0	"ABCG2 phenotype was also associated with voriconazole concentrations (p = 0.015) with poor function; (PF) or decreased function (DF) patients having higher voriconazole concentrations compared to normal function (NF) patients."	: a recom a recom d recom ole tions a recom a recom a recom

rs2231142	ABCG2	diazepam	Metabolism/PK	3	141313677	G	T	0/0	effect was seen for both Cmax and t1/2 with t-test but was not significant after Bonferroni correction.	:	

rs2231142	ABCG2	eltrombopag	Metabolism/PK		3	141313677	G	T	0/0	"The clearance of the variant of ABCG2 (rs2231142, G>T) was significantly higher than that of the wild-type (P = .02) (Figure 2A). However, when clearances were normalized by weight, the clearance between the wild-type and variant shows no statistical difference (Figure 2D). No other remarkable difference was noted in the wild-type and variant of CYP1A2 (rs762551, C > A) (Figure 2B,E) and UGT1A1*6 (rs4148323, G>A) (Figure 2C,F)."	:: Metab Clinica signific no.

rs2231142	ABCG2	imatinib	Efficacy	yes	3	141313677	G	T	0/0	"Patients with a T allele for ABCB1 (rs1045642, rs2032582, and rs1128503), A allele for ABCG2-rs2231 and G allele for CYP3A5-rs7767 polymorphisms showed better cytogenetic response and molecular response." Alleles complemented.	Synop Varian rs2231 in gene ABCG affects 42, spor 2, imatini P), heno Efficac Clinica signific 23, 2311
rs2231142	ABCG2	clozapine	Metabolism/PK	yes	3	141313677	G	T	0/0	Alleles complemented to plus chromosomal strand.	: rs2231 in gene ABCG affects respor to clozap

rs2231142	ABCG2	allopurinol	Efficacy	yes	3	141313677	G	T	0/0	Response to allopurinol was determined by whether serum uric acid concentrations decreased following allopurinol treatment. This SNP is in perfect LD with rs45499402.	Synop Varian rs2231 in gene ABCG affects respon to allopu Pheno Efficac Clinica signifi yes.

rs2231142	ABCG2	rivaroxaban	Metabolism/PK	yes	3	141313677	G	T	0/0	Alleles complemented. "In our dataset, individuals with the ABCG2 421 A/A genotype had considerably higher average values of Vd/F (508.27±72.79 L; P=0.001) and t1/2 (41.04±23.73 h; P=0.000) compared to those with the other genotypes, 421 C/A and 421 C/C, with no statistically significant difference between means." "Individuals carrying two mutant alleles in the ABCG2 SNP tended to have lower AUC, Cmax, and Cl/F (as shown in Table 4) compared to carriers of the wild-type genotype. However, it is essential to note that these differences did not achieve statistical significance (P>0.05)." This was reported for the fasting group which had 2 TT individuals, the fed group had no TT individuals.	:: Metab Clinica signific yes.

rs2231142	ABCG2	gefitinib	Efficacy	no	3	141313677	G	T	0/0	"our meta-analysis provides strong evidence that rs2231142 does not influence the response and toxicity to gefitinib treatment in patients with EGFR-mutated NSCLC." "Three studies analyzed the curative effect indicator, involving 239 patients No association was observed between the ABCG2 C421A polymorphism and the response to Gefitinib chemotherapy (HR=1.0670; 95%CI= 0.8042-1.4156; p=0.653; I2=0%), as depicted in Figure 2." Alleles complemented	Synop Varian rs2231 in gene ABCG affects respor to gefitini

rs2231142	ABCG2	rosuvastatin	Metabolism/PK	not stated	3	141313677	G	T	0/0	The authors compared PK parameters of rosuvastatin between a Caucasian group (22) a Chinese (17), Filipino (19), Indian (16), Korean (22), Vietnamese(16), and Japanese (26) groups as well as a pooled "Asian" group. AUC(0-t) and C.max were higher in subjects with the GT or TT genotypes compared with the GG genotype. The T allele may account for some but not all of the observed differences in rosuvastatin PK. Within the Asian group the T allele appeared to have a greater effect in homozygotes although only five patients in the pooled Asian group were TT while there were no TT in the Caucasian population. Geometric mean Cmax pooled Asian group TT =16.56 versus GT =28.14 and GG =17.48 and Cmax Caucasian GT =11.97 versus GG =11.56. Geometric mean AUC (0-t) pooled Asian group TT =367.8, GT =372.4, GG =199.1 and AUC(0-t) Caucasian group GT =129.9 versus TT =131.4.	: Metab Clinica signific not stated

rs2231142	ABCG2	rosuvastatin	Metabolism/PK	yes	3	141313677	G	T	0/0	The exposure to rosuvastatin increased by 44% in young subjects (p = 0.0021) with BCRP intermediate function (IF) and by 35% and 59% (p > 0.05 for both) in elderly subjects with BCRP IF and low function. The ABCG2 421C > A polymorphism was identified as a more important determinant than the SLCO1B1 521T > C polymorphism in both elderly and young subjects.	:: Metab Clinica signific yes.

rs2231142	ABCG2	imatinib	Efficacy	yes	3	141313677	G	T	0/0	Alleles complemented. There were no TT homozygotes. "However, a statistically significant difference was noted between the two patient groups regarding the distribution of different genotypes of the ABCG2 C421A polymorphism with predominance of the CA genotype in responder patients (p = 0.0395) (Fig. 2b).; Assessment of molecular response to imatinib based on the BCR-ABL1 transcript level at 12 months. Responders (n = 26); Non-responders (n = 24). "	Synop Varian rs2231 in gene ABCG affects respor to imatini Pheno Efficac Clinica signific yes.