

## PharmVIP analysis

### Project details

**Project title:** test  
**Analysis date:** 17 August 2021 4:08 p.m.  
**Analyzed by:** Hello World

### Options & parameters

**Guideline:** 17 genes ( CACNA1S, CFTR, CYP2B6, CYP2C19, CYP2C9, CYP2D6, CYP3A5, CYP4F2, DPYD, G6PD, IFNL3, NUDT15, RYR1, SLCO1B1, TPMT, UGT1A1, VKORC1 )

**HLA:** All HLA genes, Cohort ethnicity ( Caucasian, Diverse, European, Han Chinese, Indian, Japanese, Korean, Thai, Unknown, Vietnamese ), p-value < 0.05

**PGx genes:** 19 genes, ( CPIC guideline 2019, ) Transcript database ( RefSeq transcripts )

**Input files:**

<b>Guideline:</b>	VCF:	1/1/input/vcf_sample.vcf.gz
	BAM:	1/1/input/bam_sample.bam
<b>HLA:</b>	BAM:	1/1/input/bam_sample.bam
<b>PGx genes:</b>	VCF:	1/1/input/vcf_sample.vcf.gz

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## Summary: Guideline

## Related CPIC guideline(s)

**CACNA1S, RYR1**

Drug name	Diplotype	Phenotype	Recommendations	Guideline strength
<a href="#">desflurane</a> , <a href="#">enflurane</a> , <a href="#">halothane</a> , <a href="#">isoflurane</a> , <a href="#">methoxyflurane</a> , <a href="#">sevoflurane</a> , <a href="#">succinylcholine</a>	Reference/Reference, Reference/Reference	N/A, N/A	Clinical findings, family history, further genetic testing and other laboratory data should guide use of halogenated volatile anesthetics or depolarizing muscle relaxants.	<b>Strong</b>

**CFTR**

Drug name	Diplotype	Phenotype	Recommendations	Guideline strength
<a href="#">ivacaftor</a>	No CPIC variants found	N/A	This guideline does not contain recommendations for this allele combination.	N/A

**CYP2B6**

Drug name	Diplotype	Phenotype	Recommendations	Guideline strength
<a href="#">efavirenz</a>	*1/*1	Normal Metabolizer	Initiate efavirenz with standard dosing (600 mg/day).	<b>Strong</b>

**CYP2C19**

Drug name	Diplotype	Phenotype	Recommendations	Guideline strength
<a href="#">citalopram</a> , <a href="#">escitalopram</a>	*1/*2	Intermediate Metabolizer	Initiate therapy with recommended starting dose.	<b>Strong</b>
<a href="#">clopidogrel</a>	*1/*2	Intermediate Metabolizer	Alternative antiplatelet therapy (if no contraindication); e.g., prasugrel, ticagrelor	Moderate
<a href="#">sertraline</a>	*1/*2	Intermediate Metabolizer	Initiate therapy with recommended starting dose.	<b>Strong</b>
<a href="#">voriconazole</a>	*1/*2	Intermediate Metabolizer	For pediatric or adult patients: initiate therapy with recommended standard of care dosing.	Moderate

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**CYP2C19, CYP2D6**

Drug name	Diplotype	Phenotype	Recommendations	Guideline strength
<a href="#">amitriptyline</a>	*1/*2, *3/*4	Intermediate Metabolizer, Poor Metabolizer	Avoid amitriptyline use. If amitriptyline is warranted, consider 50% reduction of recommended starting dose.	Optional
<a href="#">clomipramine</a>	*1/*2, *3/*4	Intermediate Metabolizer, Poor Metabolizer	Avoid amitriptyline use. If amitriptyline is warranted, consider 50% reduction of recommended starting dose.	Optional
<a href="#">doxepin</a>	*1/*2, *3/*4	Intermediate Metabolizer, Poor Metabolizer	Avoid amitriptyline use. If amitriptyline is warranted, consider 50% reduction of recommended starting dose.	Optional
<a href="#">imipramine</a>	*1/*2, *3/*4	Intermediate Metabolizer, Poor Metabolizer	Avoid amitriptyline use. If amitriptyline is warranted, consider 50% reduction of recommended starting dose.	Optional
<a href="#">trimipramine</a>	*1/*2, *3/*4	Intermediate Metabolizer, Poor Metabolizer	Avoid amitriptyline use. If amitriptyline is warranted, consider 50% reduction of recommended starting dose.	Optional

**CYP2C9, CYP4F2, VKORC1**

Drug name	Diplotype	Phenotype	Recommendations	Guideline strength
<a href="#">warfarin</a>	*1/*2, *1/*1, No info	N/A, N/A, N/A	No Guideline.	N/A

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**CYP2D6**

Drug name	Diplotype	Phenotype	Recommendations	Guideline strength
<a href="#">atomoxetine</a>	*3/*4	Poor Metabolizer	<p><b>Children:</b> Initiate with a dose of 0.5 mg/kg/day and if no clinical response and in the absence of adverse events after 2 weeks, consider obtaining a plasma concentration 4 h after dosing. If response is inadequate and concentration is &lt;200 ng/ml, consider a proportional dose increase to achieve a concentration to approach 400 ng/ml. If unacceptable side effects are present at any time, consider a reduction in dose.</p> <p><b>Strength of recommendation:</b> Strong</p> <p><b>Adults:</b> Initiate with a dose of 40 mg/day and if no clinical response and in the absence of adverse events after 2 weeks increase dose to 80 mg/day. If response is inadequate after 2 weeks consider obtaining a plasma concentration 2-4 h after dosing. If concentration is &lt;200 ng/ml, consider a proportional dose increase to achieve a concentration to approach 400 ng/ml. If unacceptable side effects are present at any time, consider a reduction in dose.</p> <p><b>Strength of recommendation:</b> Moderate</p>	Moderate
<a href="#">codeine</a>	*3/*4	Poor Metabolizer	<p>Avoid codeine use due to lack of efficacy.</p>	Strong

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**CYP2D6**

Drug name	Diplotype	Phenotype	Recommendations	Guideline strength
<a href="#">desipramine</a>	*3/*4	Poor Metabolizer	Avoid tricyclic use due to potential for side effects. Consider alternative drug not metabolized by CYP2D6. If a TCA is warranted, consider 50% reduction of recommended starting dose.g Utilize therapeutic drug monitoring to guide dose adjustments.	Optional
<a href="#">fluvoxamine</a>	*3/*4	Poor Metabolizer	Consider a 25-50% reduction of recommended starting dose and titrate to response or use an alternative drug not metabolized by CYP2D6.	Optional
<a href="#">nortriptyline</a>	*3/*4	Poor Metabolizer	Avoid tricyclic use due to potential for side effects. Consider alternative drug not metabolized by CYP2D6. If a TCA is warranted, consider 50% reduction of recommended starting dose.g Utilize therapeutic drug monitoring to guide dose adjustments.	<b>Strong</b>
<a href="#">ondansetron</a>	*3/*4	Poor Metabolizer	Insufficient evidence demonstrating clinical impact based on CYP2D6 genotype. Initiate therapy with recommended starting dose.	No recommendation
<a href="#">paroxetine</a>	*3/*4	Poor Metabolizer	Select alternative drug not predominantly metabolized by CYP2D6 or if paroxetine use warranted, consider a 50% reduction of recommended starting dose and titrate to response.	Optional

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**CYP2D6**

Drug name	Diplotype	Phenotype	Recommendations	Guideline strength
<a href="#">tamoxifen</a>	*3/*4	Poor Metabolizer	Recommend alternative hormonal therapy such as an aromatase inhibitor for postmenopausal women or aromatase inhibitor along with ovarian function suppression in premenopausal women given that these approaches are superior to tamoxifen regardless of CYP2D6 genotype [Articles: <a href="#">26211827</a> , <a href="#">24881463</a> ] and based on knowledge that CYP2D6 poor metabolizers switched from tamoxifen to anastrozole do not have an increased risk of recurrence [Article: <a href="#">23213055</a> ]. Note, higher dose tamoxifen (40 mg/day) increases but does not normalize endoxifen concentrations and can be considered if there are contraindications to aromatase inhibitor therapy [Articles: <a href="#">27226358</a> , <a href="#">21768473</a> ].	Strong
<a href="#">tropisetron</a>	*3/*4	Poor Metabolizer	Insufficient evidence demonstrating clinical impact based on CYP2D6 genotype. Initiate therapy with recommended starting dose.	No recommendation

**CYP3A5**

Drug name	Diplotype	Phenotype	Recommendations	Guideline strength
<a href="#">tacrolimus</a>	*3/*3	Poor Metabolizer	Initiate therapy with standard recommended dose. Use therapeutic drug monitoring to guide dose adjustments.	Strong

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**DPYD**

Drug name	Diplotype	Phenotype	Recommendations	Guideline strength
<a href="#">capecitabine</a>	No CPIC decreased or no function variant with strong or moderate evidence found	Normal Metabolizer	Based on genotype, there is no indication to change dose or therapy. Use label-recommended dosage and administration.	Strong
<a href="#">fluorouracil</a>	No CPIC decreased or no function variant with strong or moderate evidence found	Normal Metabolizer	Based on genotype, there is no indication to change dose or therapy. Use label-recommended dosage and administration.	Strong

**G6PD**

Drug name	Diplotype	Phenotype	Recommendations	Guideline strength
<a href="#">rasburicase</a>	B (wildtype)/B (wildtype)	N/A	No reason to withhold rasburicase based on G6PD status.	Strong

**IFNL3**

Drug name	Diplotype	Phenotype	Recommendations	Guideline strength
<a href="#">peginterferon-alfa-2a,</a> <a href="#">peginterferon-alfa-2b,</a> <a href="#">ribavirin</a>	No info	N/A	No Guideline.	N/A

**NUDT15, TPMT**

Drug name	Diplotype	Phenotype	Recommendations	Guideline strength
<a href="#">azathioprine,</a> <a href="#">mercaptopurine</a>	*1/*1, *1/*1	N/A, N/A	No Guideline.	N/A

**SLCO1B1**

Drug name	Diplotype	Phenotype	Recommendations	Guideline strength
<a href="#">simvastatin</a>	rs4149056C/rs4149056T	N/A	Prescribe a lower dose or consider an alternative statin (e.g., pravastatin or rosuvastatin); consider routine creatine kinase (CK) surveillance	Strong

**UGT1A1**

Drug name	Diplotype	Phenotype	Recommendations	Guideline strength
<a href="#">atazanavir</a>	*1/*1	Normal Metabolizer	There is no need to avoid prescribing of atazanavir based on <i>UGT1A1</i> genetic test result.	Strong

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## Summary: HLA

## Related CPIC guideline(s)

CYP2C9, HLA-B

Drug name	Diplotype	Recommendations	Guideline strength
<a href="#">phenytoin</a>	*1/*2, Other/Other	Consider 25% reduction of recommended starting maintenance dosed (based on patient's clinical characteristics). Subsequent maintenance doses should be adjusted according to therapeutic drug monitoring and response.	Moderate

HLA-A, HLA-B

Drug name	Diplotype	Recommendations	Guideline strength
<a href="#">carbamazepine</a>	Other/Other, Other/Other	Use carbamazepine per standard dosing guidelines	Strong

HLA-B

Drug name	Diplotype	Recommendations	Guideline strength
<a href="#">abacavir</a>	Other/Other	Use abacavir per standard dosing guidelines	Strong
<a href="#">allopurinol</a>	Other/Other	Use allopurinol per standard dosing guidelines.	Strong
<a href="#">oxcarbazepine</a>	Other/Other	Use oxcarbazepine per standard dosing guidelines	Strong

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## Related publication(s) for HLA-ADR association

## HLA-A\*01 (No. of detecting tools = 3/3)

Drug name	ADR	Patient disease	Cohort ethnicity	No. of pub.(s)
Clozapine	agranulocytosis	Schizophrenia	Caucasian	1

## HLA-A\*01:01 (No. of detecting tools = 3/3)

Drug name	ADR	Patient disease	Cohort ethnicity	No. of pub.(s)
Amoxicillin-clavulanate	drug induced liver injury (DILI)	Bacterial infections	Caucasian	2
Antibacterial drugs	Stevens–Johnson syndrome (SJS),toxic epidermal necrolysis (TEN),drug-induced hypersensitivity syndrome (DIHS),acute generalized exanthematous pustulosis (AGEP)	Bacterial infections	Unknown	2
Antiepileptic drugs	Stevens-Johnson syndrome (SJS),toxic epidermal necrolysis (TEN)	Epilepsy	Han Chinese	1
Antiepileptic drugs	Stevens–Johnson syndrome (SJS),toxic epidermal necrolysis (TEN),drug-induced hypersensitivity syndrome (DIHS),acute generalized exanthematous pustulosis (AGEP)	Epilepsy	Unknown	1
Carbamazepine	Stevens-Johnson syndrome (SJS),toxic epidermal necrolysis (TEN),drug-induced hypersensitivity syndrome (DIHS)	Epilepsy	Japanese	1
Carbamazepine	Stevens-Johnson syndrome (SJS),toxic epidermal necrolysis (TEN)	Epilepsy	Han Chinese	2
Carbamazepine	Stevens-Johnson syndrome (SJS),toxic epidermal necrolysis (TEN)	Epilepsy	European	2
EGF receptor inhibitors	maculopapular exanthema (MPE)	Cancer	Unknown	1
Lamotrigine	Stevens–Johnson syndrome (SJS),toxic epidermal necrolysis (TEN)	Epilepsy	Caucasian	1
Lamotrigine	drug-induced hypersensitivity syndrome (DIHS)	Epilepsy	Caucasian	1
Levetiracetam	maculopapular eruption (MPE)	Epilepsy	Indian	1
NSAID and 'multi-ingredient cold medication'	Stevens-Johnson syndrome (SJS),toxic epidermal necrolysis (TEN),severe ocular complications	Common cold	Diverse	1

## HLA-A\*11 (No. of detecting tools = 3/3)

Drug name	ADR	Patient disease	Cohort ethnicity	No. of pub.(s)
Tetanus antitoxin	maculopapular eruption (MPE)	Tetanus	Han Chinese	1

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## HLA-A\*11:01 (No. of detecting tools = 2/3)

Drug name	ADR	Patient disease	Cohort ethnicity	No. of pub.(s)
Amoxicillin-clavulanate	drug induced liver injury (DILI)	Bacterial infections	Caucasian	2
Antibacterial drugs	Stevens–Johnson syndrome (SJS),toxic epidermal necrolysis (TEN),drug-induced hypersensitivity syndrome (DIHS),acute generalized exanthematous pustulosis (AGEP)	Bacterial infections	Unknown	2
Antiepileptic drugs	Stevens–Johnson syndrome (SJS),toxic epidermal necrolysis (TEN),drug-induced hypersensitivity syndrome (DIHS),acute generalized exanthematous pustulosis (AGEP)	Epilepsy	Unknown	1
Carbamazepine	Stevens–Johnson syndrome (SJS),toxic epidermal necrolysis (TEN)	Epilepsy	Caucasian	1
Carbamazepine	Stevens-Johnson syndrome (SJS),toxic epidermal necrolysis (TEN),drug-induced hypersensitivity syndrome (DIHS)	Epilepsy	Japanese	1
Carbamazepine	Stevens-Johnson syndrome (SJS),toxic epidermal necrolysis (TEN)	Epilepsy	Han Chinese	4
Carbamazepine	drug-induced hypersensitivity syndrome (DIHS)	Epilepsy	Han Chinese	2
Carbamazepine	maculopapular exanthema (MPE)	Epilepsy	Han Chinese	3
Clarithromycin	drug-induced skin reaction (DISI)	Bacterial infections	Han Chinese	1
Lamotrigine	maculopapular exanthema (MPE)	Epilepsy	Han Chinese	1
Lamotrigine	Stevens–Johnson syndrome (SJS),toxic epidermal necrolysis (TEN)	Epilepsy	Caucasian	1
Levetiracetam	maculopapular eruption (MPE)	Epilepsy	Indian	1
NSAID and 'multi-ingredient cold medication'	Stevens-Johnson syndrome (SJS),toxic epidermal necrolysis (TEN),severe ocular complications	Common cold	Diverse	1
Phenytoin	Stevens–Johnson syndrome (SJS),toxic epidermal necrolysis (TEN)	Epilepsy	Caucasian	1
Phenytoin	Stevens-Johnson syndrome (SJS),toxic epidermal necrolysis (TEN)	Epilepsy	Han Chinese	1
Tetanus antitoxin	maculopapular eruption (MPE)	Tetanus	Han Chinese	1

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**HLA-B\*08:01 (No. of detecting tools = 3/3)**

Drug name	ADR	Patient disease	Cohort ethnicity	No. of pub.(s)
Amoxicillin-clavulanate	drug induced liver injury (DILI)	Bacterial infections	Caucasian	2
Antibacterial drugs	Stevens–Johnson syndrome (SJS),toxic epidermal necrolysis (TEN),drug-induced hypersensitivity syndrome (DIHS),acute generalized exanthematous pustulosis (AGEP)	Bacterial infections	Unknown	2
Antiepileptic drugs	Stevens–Johnson syndrome (SJS),toxic epidermal necrolysis (TEN),drug-induced hypersensitivity syndrome (DIHS),acute generalized exanthematous pustulosis (AGEP)	Epilepsy	Unknown	1
Carbamazepine	Stevens-Johnson syndrome (SJS),toxic epidermal necrolysis (TEN)	Epilepsy	European	2
Carbamazepine	Stevens-Johnson syndrome (SJS),toxic epidermal necrolysis (TEN),drug-induced hypersensitivity syndrome (DIHS)	Epilepsy	Vietnamese	1
EGF receptor inhibitors	maculopapular exanthema (MPE)	Cancer	Unknown	1
Lamotrigine	drug-induced hypersensitivity syndrome (DIHS)	Epilepsy	Caucasian	1
NSAID and 'multi-ingredient cold medication'	Stevens-Johnson syndrome (SJS),toxic epidermal necrolysis (TEN),severe ocular complications	Common cold	Diverse	1
Phenytoin	Stevens–Johnson syndrome (SJS),toxic epidermal necrolysis (TEN)	Epilepsy	Caucasian	1

**HLA-B\*56:01 (No. of detecting tools = 3/3)**

Drug name	ADR	Patient disease	Cohort ethnicity	No. of pub.(s)
Amoxicillin-clavulanate	drug induced liver injury (DILI)	Bacterial infections	Caucasian	2
Antibacterial drugs	Stevens–Johnson syndrome (SJS),toxic epidermal necrolysis (TEN),drug-induced hypersensitivity syndrome (DIHS),acute generalized exanthematous pustulosis (AGEP)	Bacterial infections	Unknown	2
Antiepileptic drugs	Stevens–Johnson syndrome (SJS),toxic epidermal necrolysis (TEN),drug-induced hypersensitivity syndrome (DIHS),acute generalized exanthematous pustulosis (AGEP)	Epilepsy	Unknown	1
Carbamazepine	Stevens-Johnson syndrome (SJS),toxic epidermal necrolysis (TEN)	Epilepsy	Thai	1

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**HLA-B\*56:01 (No. of detecting tools = 3/3)**

Drug name	ADR	Patient disease	Cohort ethnicity	No. of pub.(s)
Carbamazepine	Stevens-Johnson syndrome (SJS),toxic epidermal necrolysis (TEN),drug-induced hypersensitivity syndrome (DIHS),maculopapular exanthema (MPE)	Epilepsy	Japanese	1
Carbamazepine	Stevens-Johnson syndrome (SJS),toxic epidermal necrolysis (TEN)	Epilepsy	Han Chinese	1
NSAID and 'multi-ingredient cold medication'	Stevens-Johnson syndrome (SJS),toxic epidermal necrolysis (TEN),severe ocular complications	Common cold	Diverse	1
Oxcarbazepine	maculopapular exanthema (MPE)	Epilepsy	Han Chinese	1
Phenytoin	Stevens-Johnson syndrome (SJS),toxic epidermal necrolysis (TEN)	Epilepsy	Han Chinese	1

**HLA-C\*01:02 (No. of detecting tools = 3/3)**

Drug name	ADR	Patient disease	Cohort ethnicity	No. of pub.(s)
Carbamazepine	drug-induced hypersensitivity syndrome (DIHS)	Epilepsy	Han Chinese	1
Carbamazepine	maculopapular exanthema (MPE)	Epilepsy	Han Chinese	1
Carbamazepine	Stevens-Johnson syndrome (SJS)	Epilepsy	Korean	1
Carbamazepine	drug-induced hypersensitivity syndrome (DIHS)	Epilepsy	Korean	1
Carbamazepine	drug-induced skin reaction (DISI)	Epilepsy	Korean	1
Carbamazepine	Stevens-Johnson syndrome (SJS),toxic epidermal necrolysis (TEN)	Epilepsy	Han Chinese	1
Clarithromycin	drug-induced skin reaction (DISI)	Bacterial infections	Han Chinese	1
Lamotrigine	maculopapular eruption (MPE)	Epilepsy	Korean	1
NSAID and 'multi-ingredient cold medication'	Stevens-Johnson syndrome (SJS),toxic epidermal necrolysis (TEN),severe ocular complications	Common cold	Diverse	1
Phenytoin	Stevens-Johnson syndrome (SJS),toxic epidermal necrolysis (TEN)	Epilepsy	Han Chinese	1
Tetanus antitoxin	maculopapular eruption (MPE)	Tetanus	Han Chinese	1

**HLA-C\*07 (No. of detecting tools = 3/3)**

Drug name	ADR	Patient disease	Cohort ethnicity	No. of pub.(s)
Clozapine	agranulocytosis	Schizophrenia	Caucasian	1

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**HLA-C\*07:01 (No. of detecting tools = 3/3)**

Drug name	ADR	Patient disease	Cohort ethnicity	No. of pub.(s)
EGF receptor inhibitors	maculopapular exanthema (MPE)	Cancer	Unknown	1
Lamotrigine	Stevens–Johnson syndrome (SJS),toxic epidermal necrolysis (TEN)	Epilepsy	Caucasian	1
Lamotrigine	drug-induced hypersensitivity syndrome (DIHS)	Epilepsy	Caucasian	1
Lamotrigine	maculopapular eruption (MPE)	Epilepsy	Korean	1
NSAID and 'multi-ingredient cold medication'	Stevens-Johnson syndrome (SJS),toxic epidermal necrolysis (TEN),severe ocular complicatoins	Common cold	Diverse	1
Phenytoin	Stevens–Johnson syndrome (SJS),toxic epidermal necrolysis (TEN)	Epilepsy	Caucasian	1
Phenytoin	drug-induced hypersensitivity syndrome (DIHS)	Epilepsy	Caucasian	1

**HLA-DPB1\*04:01 (No. of detecting tools = 3/3)**

Drug name	ADR	Patient disease	Cohort ethnicity	No. of pub.(s)
Bucillamine	bucillamine-induced proteinuria	Rheumatoid arthritis	Japanese	1

**HLA-DPB1\*14:01 (No. of detecting tools = 3/3)**

Drug name	ADR	Patient disease	Cohort ethnicity	No. of pub.(s)
Bucillamine	bucillamine-induced proteinuria	Rheumatoid arthritis	Japanese	1

**HLA-DQA1\*01:01 (No. of detecting tools = 3/3)**

Drug name	ADR	Patient disease	Cohort ethnicity	No. of pub.(s)
Amoxicillin-clavulanate	drug induced liver injury (DILI)	Bacterial infections	Caucasian	2

**HLA-DQA1\*05:01 (No. of detecting tools = 3/3)**

Drug name	ADR	Patient disease	Cohort ethnicity	No. of pub.(s)
Amoxicillin-clavulanate	drug induced liver injury (DILI)	Bacterial infections	Caucasian	2

**HLA-DQB1\*02:01 (No. of detecting tools = 3/3)**

Drug name	ADR	Patient disease	Cohort ethnicity	No. of pub.(s)
Amoxicillin-clavulanate	drug induced liver injury (DILI)	Bacterial infections	Caucasian	2
Clozapine	agranulocytosis	Schizophrenia	Caucasian	1

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**HLA-DQB1\*05:01 (No. of detecting tools = 3/3)**

Drug name	ADR	Patient disease	Cohort ethnicity	No. of pub.(s)
Amoxicillin-clavulanate	drug induced liver injury (DILI)	Bacterial infections	Caucasian	2
Bucillamine	bucillamine-induced proteinuria	Rheumatoid arthritis	Japanese	1
Oxcarbazepine	maculopapular exanthema (MPE)	Epilepsy	Korean	1

**HLA-DRB1\*01 (No. of detecting tools = 3/3)**

Drug name	ADR	Patient disease	Cohort ethnicity	No. of pub.(s)
Flucloxacillin	drug induced liver injury (DILI)	Bacterial infections	Caucasian	1

**HLA-DRB1\*01:01 (No. of detecting tools = 3/3)**

Drug name	ADR	Patient disease	Cohort ethnicity	No. of pub.(s)
Amoxicillin-clavulanate	drug induced liver injury (DILI)	Bacterial infections	Caucasian	2
Antiepileptic drugs	Stevens-Johnson syndrome (SJS),toxic epidermal necrolysis (TEN)	Epilepsy	Han Chinese	1
Bucillamine	bucillamine-induced proteinuria	Rheumatoid arthritis	Japanese	1
Carbamazepine	maculopapular exanthema (MPE)	Epilepsy	Han Chinese	1
Carbamazepine	Stevens-Johnson syndrome (SJS),toxic epidermal necrolysis (TEN)	Epilepsy	Han Chinese	1
Clozapine	agranulocytosis	Schizophrenia	Caucasian	1
Co-trimoxazole	maculopapular exanthema (MPE),Stevens-Johnson syndrome (SJS)	HIV	Caucasian	1
EGF receptor inhibitors	maculopapular exanthema (MPE)	Cancer	Unknown	1
Nevirapine	drug-induced hypersensitivity syndrome (DIHS)	HIV	Diverse	1
Nevirapine	drug-induced skin reaction (DISI),drug induced liver injury (DILI)	HIV	Caucasian	1

**HLA-DRB1\*03 (No. of detecting tools = 3/3)**

Drug name	ADR	Patient disease	Cohort ethnicity	No. of pub.(s)
Flucloxacillin	drug induced liver injury (DILI)	Bacterial infections	Caucasian	1

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**HLA-DRB1\*03:01 (No. of detecting tools = 3/3)**

Drug name	ADR	Patient disease	Cohort ethnicity	No. of pub.(s)
Allopurinol	Stevens-Johnson syndrome (SJS),toxic epidermal necrolysis (TEN),drug-induced hypersensitivity syndrome (DIHS)	Gout	Han Chinese	1
Amoxicillin-clavulanate	drug induced liver injury (DILI)	Bacterial infections	Caucasian	2
Carbamazepine	maculopapular exanthema (MPE)	Epilepsy	Han Chinese	1
Clozapine	agranulocytosis	Schizophrenia	Caucasian	1
Co-trimoxazole	maculopapular exanthema (MPE),Stevens-Johnson syndrome (SJS)	HIV	Caucasian	1
Nevirapine	drug-induced hypersensitivity syndrome (DIHS)	HIV	Diverse	1
Phenytoin	Stevens-Johnson syndrome (SJS),toxic epidermal necrolysis (TEN)	Epilepsy	Han Chinese	1

**HLA-DRB3\*01:01 (No. of detecting tools = 3/3)**

Drug name	ADR	Patient disease	Cohort ethnicity	No. of pub.(s)
Clozapine	agranulocytosis	Schizophrenia	Caucasian	1

**CONTACT**

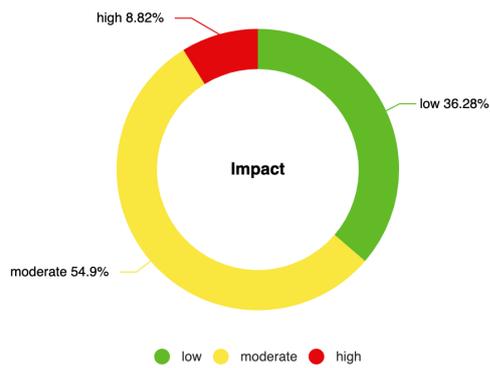
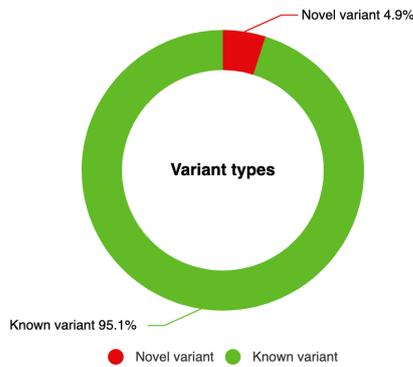
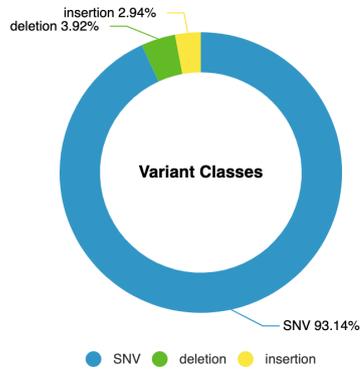
 National Biobank of Thailand (NBT)

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## Summary: PGx genes



**Number of variants categorized by classes**

Variant classes	# variants
SNV	95
deletion	4
insertion	3

**Number of variants categorized by types**

Type	# variants
Novel variation	5
Known variant	97

**Number of transcripts categorized by variant impacts**

Impacts	# transcripts
low	37
moderate	56
high	9

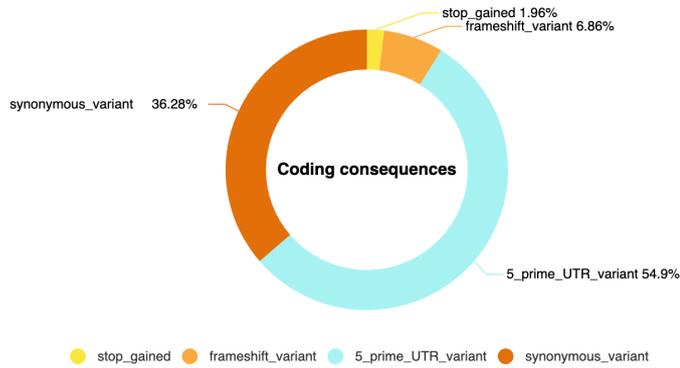
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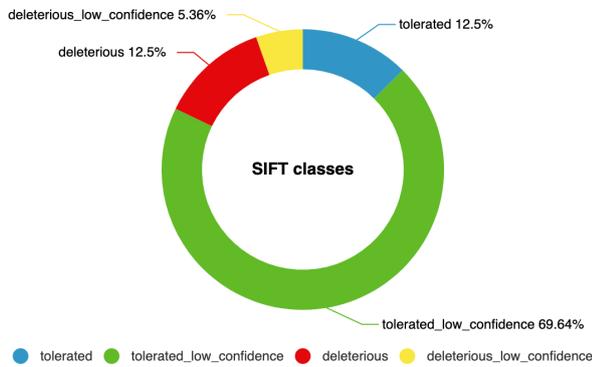
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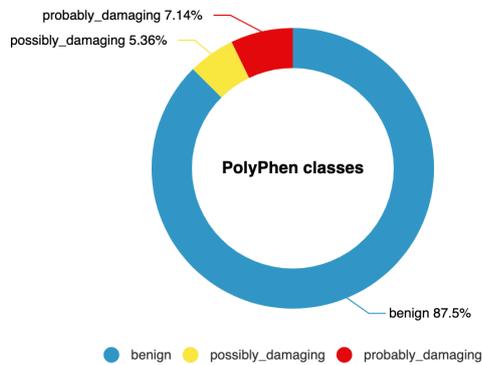




stop\_gained frameshift\_variant 5\_prime\_UTR\_variant synonymous\_variant



tolerated tolerated\_low\_confidence deleterious deleterious\_low\_confidence



benign possibly\_damaging probably\_damaging

### Number of transcripts categorized by coding consequences

Coding consequences	# transcripts
Stop gained	2
Frameshift variant	7
Missense variant	56
Splice region variant	5
Synonymous variant	37

### Number of transcripts categorized by SIFT classes

SIFT classes	# transcripts
tolerated	7
tolerated_low_confidence	39
deleterious	7
deleterious_low_confidence	3

### Number of transcripts categorized by PolyPhen classes

PolyPhen classes	# transcripts
benign	49
possibly_damaging	3
probably_damaging	4

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