



有償アドオン「VSPGx」は、薬理ゲノミクスにおける臨床的解釈を行うためのソフトウェアです。このアドオンで利用可能なPGx Variant Detection and Recommendationアルゴリズムは、CYPなどの薬物代謝酵素の遺伝子におけるディプロタイプを特定し、それらにClinical Pharmacogenetics Implementation Consortium (CPIC)によって推奨されている薬剤情報のアノテーションを付けます。

CYP2D6 Diplotype	Metabolizer Phenotype
CYP2D6 *1/*1	Normal metabolizer
CYP2D6 *2/*122	Intermediate metabolizer
CYP2D6 *3/*3	Poor Metabolizer
CYP2D6 *1/*1x2	Ultrarapid metabolizer
CYP2D6 *5/*5	Poor metabolizer

- Required Variants for CYP2D6 *2/*122
- CYP2D6 *2: 2851C>T (rs16947), 4181G>C (rs1135840)
 - CYP2D6*122 3280G>A (rs61745683)

Table 2 Codeine therapy recommendations based on CYP2D6 phenotype

Phenotype	Activity score	Implications	Recommendations	Classification of recommendation*
CYP2D6 ultrarapid metabolizer	> 2.25	Increased formation of morphine leading to higher risk of toxicity	Avoid codeine use because of potential for serious toxicity. If opioid use is warranted, consider a non-tramadol opioid.	Strong
CYP2D6 normal metabolizer	1.25 ≤ x ≤ 2.25	Expected morphine formation	Use codeine label recommended age-specific or weight-specific dosing.	Strong
CYP2D6 intermediate metabolizer	0 < x < 1.25	Reduced morphine formation	Use codeine label recommended age-specific or weight-specific dosing. If no response and opioid use is warranted, consider a non-tramadol opioid.	Moderate
CYP2D6 poor metabolizer	0	Greatly reduced morphine formation leading to diminished analgesia.	Avoid codeine use because of possibility of diminished analgesia. If opioid use is warranted, consider a non-tramadol opioid.	Strong
CYP2D6 indeterminate	n/a	n/a	No recommendation	No recommendation

n/a, not applicable.
*Rating scheme described in the **Supplementary Material**.

Golden Labs Precision Medicine

Patient Name: Jane Doe | Report Date: 02/27/2024

Current Patient Medications

List of provided patient medications with recommendations:

Sertraline (CPIC Level A Gene-Drug)
 CYP2C19: Normal Metabolizer
 CYP2B6: Poor Metabolizer
 Consider a lower starting dose, slower titration schedule and 25% reduction of standard maintenance dose as compared to CYP2B6 normal metabolizers or select a clinically appropriate alternative antidepressant not predominantly metabolized by CYP2B6.
 CYP2D6, CYP2C19, CYP2B6, SLCO4A4, HTR2A and Serotonin Reuptake Inhibitor Antidepressants (<https://cpicpgx.org/guidelines/cpic-guideline-for-srri-and-srni-antidepressants/>)

Pharmacogenomic Results Summary

CATEGORY	DRUG CLASS	MAJOR GENE-DRUG INTERACTIONS	MODERATE GENE-DRUG INTERACTIONS	MINIMAL GENE-DRUG INTERACTIONS
Cardiovascular	Antiplatelets			Clopidogrel
	Statins			Fluvastatin Atorvastatin Lovastatin Pitavastatin Pravastatin Rosuvastatin Simvastatin
Gastrointestinal	Proton Pump Inhibitors			Dexlansoprazole Omeprazole Lansoprazole Pantoprazole
Infections	Anti-HIV Agents	Atazanavir	Efavirenz	
	Antifungals			Voriconazole
Misc	Analgesics			Lornoxicam Tenoxicam
Pain	NSAIDs			Flurbiprofen Celecoxib

【アレルの同定】

CPICガイドラインに従い、薬物代謝酵素などの遺伝子上に存在するスターアレルを特定し、遺伝子のディプロタイプを同定します。

【推奨情報とのマッチング】

各遺伝子にディプロタイプが割り当てられると、VSPGxはこれらのディプロタイプと対応する表現型および薬剤投与などの推奨事項と照合します。

【構造多型】

PGx Variant Detection and Recommendationアルゴリズムを使用して、構造的多型およびコピー数の欠失または重複と関連する表現型データと治療推奨事項をレポートすることができます。

【レポート出力】

PGx Variant Detection and Recommendationアルゴリズムを実行した後、VSPGxのカスタマイズ可能なレポートシステムを使用して臨床レポートを作成できます。VSPGxには、カスタムレポート作成に便利なMicrosoft Wordベースのレポートテンプレートが付属しています。

Variants: 183 | PGx Genes: 9

Filter Variants: PGX-001

PGx Gene Info	Gene	Diploype	Phenotype	PGX-001	CPIC Must Call Varia...	PGx Annotations	PGx Annotations for PGX-001
Region	UGT1A1	*1/*80+*28	Intermediate Metabolizer				
2-233755270-233778300	UGT1A1	*1/*80+*28	Intermediate Metabolizer				
7-99643194-99684996	CYP3A5	*3/*3	Poor Metabolizer				
10-94757681-94860547	CYP2C19	*35/*35	Poor Metabolizer				
10-94933658-94995091	CYP2C9	*1/*1	Normal Metabolizer				
12-21126194-21244796	SLCO1B1	*1/*1	Normal Function				
16-31085854-31099797	VKORC1	rs9923231 reference ...	?				
19-15873023-15903074	CYP4F2	*1/*1	?				
19-40986282-41023398	CYP2B6	*1/*1	Normal Metabolizer				
22-42121499-42135810	CYP2D6	*2/*122	Indeterminate				