

# ePGA report

This is the eMoDiA report for the phase 3 sample HG0096 of 1000 Genomes Project

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## ePGA: Purpose and Use

ePGA TRANSLATION SERVICE IS SOLELY FOR RESEARCH, DEMONSTRATIONAL & EDUCATIONAL PURPOSES

ePGA Translation Service is meant to DEMONSTRATE the **potential** of translating validated PGx knowledge into clinical practice

In its current implementation ePGA Translation Service use just public-domain and completely anonymous genotype profiles from the 1000 Genomes project (Phase I/1092 sample cases, Phase III / 2504 sample cases) in order to **demonstrate** its functionality

- The ePGA Translation Service adheres to the emerging trend of pre-emptive high-throughput genotyping as a vital clinical (diagnostic and prognostic) decision making component aiming to offer respective genotype-to-phenotype inference services as a mean to translate PGx knowledge from bench-to-bedside.
  - The ePGA Translation Service is based on ‘matching’ individual genotype (SNP) profiles with PGx gene haplotypes, and the subsequent inference of the corresponding metabolizer phenotypes. Currently ePGA/Translation component employs harmonized haplotypes-tables as registered and curated by [PharmGKB](#) (the most advanced PGx knowledge base). The developers of ePGA are planning to expand the system in order to capture and cover PGx knowledge from other sources as well.
  - ePGA developers reserve the right at any time, to make changes to the whole or any part of the services offered on this Web-site as it deems appropriate.
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sample	gene	diplootype	phenotype
HG00096	ABCC2	H15/H5	Var/Var
HG00096	ADRB1	H1/H1	WT/WT
HG00096	ADRB2	4.0/4.0	Var/Var
HG00096	APOE	E3/E3	WT/WT
HG00096	CDA	1A/1A	WT/WT
HG00096	CFTR	Reference/Reference	WT/WT
HG00096	CHRNA5	haplotype 1/haplotype 3	WT/Var
HG00096	COMT	Haplotype high activity/Haplotype intermediate activity	WT/Var
HG00096	CYP1A1	1/1	WT/WT
HG00096	CYP1A2	1M/1M	Var/Var
HG00096	CYP1B1	1/4	WT/Var
HG00096	CYP2A13	1/1	WT/WT
HG00096	CYP2A6	1A/1A	WT/WT
HG00096	CYP2C8	1A/4	WT/Var
HG00096	CYP2C9	1/1	WT/WT
HG00096	CYP2E1	1/1	WT/WT
HG00096	CYP2R1	1/1	WT/WT

sample	gene	diplotype	phenotype
HG00096	CYP2S1	1A/1A	WT/WT
HG00096	CYP2W1	1A/1A	WT/WT
HG00096	CYP3A43	1/1	WT/WT
HG00096	CYP3A7	2/2	Var/Var
HG00096	CYP4B1	1/2A	WT/Var
HG00096	CYP4F2	1/1	WT/WT
HG00096	DDC	#1/#3	WT/Var
HG00096	DPYD	1/9A	WT/Var
HG00096	DPYD_2	rs67376798T/rs67376798T	WT/WT
HG00096	G6PD	B (wildtype)/B (wildtype)	WT/WT
HG00096	HMGCR	H2/H2	WT/WT
HG00096	HTR2C	1-2-1/2-1-1	WT/Var
HG00096	IGFBP3	7/10	Var/Var
HG00096	LDLR	L1/L1	Var/Var
HG00096	NAT1	4/10	WT/Var
HG00096	P2RY12_2	H1/H2	WT/Var
HG00096	PIK3CA	H1/H3	WT/Var
HG00096	SCN1A	#1/#2	WT/Var
HG00096	SCN5A	Haplotype A/Haplotype A	WT/WT
HG00096	SCNN1B	1_1/1_2	WT/Var
HG00096	SLC22A1	1/1	WT/WT
HG00096	SLC25A27	CCAC/CTAT	WT/Var
HG00096	SULT1A2	1/1	WT/WT
HG00096	SULT1C2	1/2	WT/Var
HG00096	SULT2A1	1/1	WT/WT
HG00096	SULT4A1	#2/#6	Var/Var
HG00096	TPMT	1/1	WT/WT
HG00096	UGT1A10	1a/1a	WT/WT
HG00096	UGT1A3	1/6	WT/Var
HG00096	UGT1A3_2	1a/6a	WT/Var
HG00096	UGT1A4	1a/1b	WT/Var
HG00096	UGT1A5	1/1	WT/WT
HG00096	UGT1A8	1a/1a	WT/WT
HG00096	UGT1A9	1a/1q	WT/Var
HG00096	UGT2B15	4/5	Var/Var