epigeneti**X** Epi23

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ACMG Incidental Findings

The American College of Medical Genetics and Genomics (ACMG) has published a list of 94 genes, for which specific mutations are known to cause disorders with established phenotypes that are clinically actionable by a widely acknowledged intervention. Below is a list of non-benign variants that have gnomAD AF <= 0.05 or missing AF and are found in these genes.

ClinVar Condition	ClinVar Significance	ACMG Ciassification	Status	gnomAD AF	Gene Name	Chr	Start	Stop	Ref	Alt

No variants detected

Other Findings - Heterozygous Variants

Below is a list of **pathogenic heterozygous** variants that are not found in the list of genes compiled by ACMG as actionable.

Gene Name	Chr	Start	Stop	Ref	Alt	gnomAD AF	ClinVar Condition	ClinVar Significance
CNN2	19	1037767	1037767	G-G	G-A	0.0000846	Pulmonary artery atresia	⚠ Pathogenic
CNN2	19	1037757	1037757	G-G	G-A	0.0001331	Pulmonary artery atresia	⚠ Pathogenic
TFAM	10	58388809	58388810	CA-CA	CA-C	0.0128700	Mitochondrial DNA depletion syndrome 15 (hepatocerebral type)	⚠ Likely pathogenic
SLC9B1	4	102901325	102901327	AAC-AAC	AAC-A	0.0036410	Usher syndrome	⚠ Pathogenic

Other Findings - Homozygous Variants

Below is a list of **pathogenic homozygous** variants that are not found in the list of genes compiled by ACMG as actionable.

Gene Name	Chr	Start	Stop	Ref	Alt	gnomAD AF	ClinVar Condition	ClinVar Significance

No variants detected

Health Predisposition

In this section, your genetic predispositions are listed. We compare 15 different genomic positions to identify 7 different possible predispositions that might you have. Any predispositions based on specific variants are investigated and reported below. Only the predispositions that you have are reported below. This test does not diagnose any health conditions. If you have any concerns that you or your family might have any condition, please contact with a healthcare professional.

Condition	Gene	RS ID	Results	Status	Clinical Explanation

No variants detected

Drug-Variant Interactions

In this section, you may observe the **toxicity**-labelled drug-variant interactions. We analyze more than 140 genomic positions to identify Drug-Variant Interactions. Only Tier-1A variants from PharmGKB database are represented. Only the variants that you have are reported below.

Gene	Chemicals	Genotype	Clinical Annotation
DPYD	capecitabine	TC	Both variants of rs1801159 are assigned normal function by CPIC. Patients with the CT genotype and cancer who are treated with capecitabine, a fluoropyrimidine-based chemotherapy, may not have altered risk of drug toxicity as compared to patients with the CC or TT genotype. However, conflicting evidence has been reported. Other genetic and clinical factors may also influence risk of drug toxicity.
DPYD	fluorouracil	TC	Both variants of rs1801159 are assigned normal function by CPIC. Patients with the CT genotype and cancer who are treated with fluorouracil, a fluoropyrimidine-based chemotherapy, may not have altered risk of drug toxicity as compared to patients with the CC or TT genotype. However, conflicting evidence has been reported. Other genetic and clinical factors may also influence risk of drug toxicity.
DPYD	capecitabine	AA	Both variants of rs1801265 are assigned normal function by CPIC. Patients with the AA genotype and cancer who are treated with capecitabine, a fluoropyrimidine-based chemotherapy, may not have altered risk of drug toxicity as compared to patients with the AG or GG genotype. However, conflicting evidence has been reported. Other genetic and clinical factors may also influence risk of drug toxicity.
DPYD	fluorouracil	AA	Both variants of rs1801265 are assigned normal function by CPIC. Patients with the AA genotype and cancer who are treated with fluorouracil, a fluoropyrimidine-based chemotherapy, may not have altered risk of drug toxicity as compared to patients with the AG or GG genotype. However, conflicting evidence has been reported. Other genetic and clinical factors may also influence risk of drug toxicity.
SLCO1B1	atorvastatin	CC	Patients with the rs4149056 CC genotype may have an increased risk of myopathy when treated with atorvastatin as compared to patients with the TT genotype. However, conflicting evidence has been reported. Other genetic and clinical factors may also influence risk of developing myopathy when treated with atorvastatin.
SLCO1B1	lovastatin	CC	Patients with the rs4149056 CC genotype may have an increased risk of lovastatin-related myopathy when treated with lovastatin as compared to patients with the TT genotype. Other genetic and clinical factors may also influence risk of toxicity to lovastatin.
SLCO1B1	rosuvastatin	CC	Patients with the rs4149056 CC genotype may have increased risk of statin-related myopathy or myalgia when treated with rosuvastatin as compared to patients with genotype TT. However, conflicting evidence has been reported. Other genetic and clinical factors may also affect risk to rosuvastatin.
SLCO1B1	pravastatin	CC	Patients with the rs4149056 CC genotype may have an increased risk of developing myopathy when treated with pravastatin as compared to patients with the TT genotype. However, conflicting evidence has been reported. Other genetic and clinical factors may also affect a patient's risk of experiencing pravastatin-induced myopathy.

Gene	Chemicals	Genotype	Clinical Annotation
SLCO1B1	simvastatin	CC	Patients with the rs4149056 CC genotype may have a higher risk of simvastatin-related myopathy when treated with simvastatin as compared to patients with the TT genotype. However, conflicting evidence has been reported. Other genetic and clinical factors may also influence risk of toxicity to simvastatin.
SLCO1B1	fluvastatin	CC	Patients with the rs4149056 CC genotype may have an increased risk of developing myopathy when treated with fluvastatin as compared to patients with the TT genotype. However, conflicting evidence has been reported. Other genetic and clinical factors may also affect a patient's risk of developing fluvastatin-induced myopathy.
RYR1	desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine	GG	Patients with the rs193922753 GG genotype may have a decreased, but not absent, risk for malignant hyperthermia based on this variant when treated with volatile anesthetics (desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane) and/or succinylcholine as compared to patients with genotype GT and TT. Other genetic or clinical factors may also influence the risk for malignant hyperthermia.
RYR1	desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine	GG	Patients with the rs121918592 GG genotype may have a decreased, but not absent, risk for malignant hyperthermia based on this variant when treated with volatile anesthetics (desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane) and/or succinylcholine as compared to patients with genotype CC, CG, AA or AG. Other genetic or clinical factors may also influence the risk for malignant hyperthermia.
RYR1	desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine	GG	Patients with the rs193922772 GG genotype may have a decreased, but not absent, risk for malignant hyperthermia based on this variant when treated with volatile anesthetics (desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane) and/or succinylcholine as compared to patients with the GT or TT genotype. Other genetic or clinical factors may also influence the risk for malignant hyperthermia.
RYR1	desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane, succinylcholine	GG	Patients with the rs63749869 GG genotype may have a decreased, but not absent, risk for malignant hyperthermia based on this variant when treated with volatile anesthetics (desflurane, enflurane, halothane, isoflurane, methoxyflurane, sevoflurane) and/or succinylcholine as compared to patients with the AG or AA genotype. Other genetic or clinical factors may also influence the risk for malignant hyperthermia.
CYP2B6	efavirenz	GT	Patients with the rs3745274 GT genotype may have an increased risk of efavirenz-induced side effects, including sleep- and central nervous system-related side effects, as compared to patients with the GG genotype. However, conflicting evidence has been reported. Other genetic and clinical factors may also influence risk of efavirenz toxicity.

Allele Functions

Gene	Allele	Function
CYP2C19	*25	Decreased function
CYP2D6	*28	Uncertain function

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