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DNA INTERPRETATION

Comprehensive Clinical Genomic Interpretation Report

PATIENT	ID	AGE	GENDER
Sample Patient	PT-SAMPLE	43	Male

GENERATED	LANGUAGE
2026-09-23 00:19:06	EN

Executive summary

This genomic evaluation incorporates the patient's context of ongoing thyroid monitoring, noting a discrepancy with the demographic data printed on the laboratory report (demographic profile lists a 43-year-old male, whereas the requisition specifies female born in 1984). Clinically critical findings include an APOE e3/e4 genotype conferring increased cardiovascular and late-onset neurodegenerative risk, an LPA risk allele associated with elevated lipoprotein(a), and heterozygous carrier status for hereditary hemochromatosis (HFE C282Y). Pharmacogenomic profiling reveals actionable variants including CYP2C19 intermediate metabolizer (*1/*2), CYP2C9 *1/*2 with VKORC1 sensitivity (affecting warfarin requirements), and SLCO1B1 *1/*5 associated with statin-induced myopathy risk. Additional nutrigenomic findings highlight lactase non-persistence, reduced folate metabolism (MTHFR C677T heterozygous), and altered vitamin D handling.

Overall assessment

Elevated Overall genomic risk is categorized as elevated, driven predominantly by cardiovascular risk alleles (APOE e3/e4, LPA, CDKN2B-AS1), actionable pharmacogenomic sensitivities (SLCO1B1, CYP2C19, CYP2C9/VKORC1), and HFE C282Y carrier status requiring biochemical iron surveillance alongside ongoing thyroid monitoring.

Data quality

Source: Saliva-based genome-wide SNP genotyping array (~650,000 markers, reference build GRCh37, plus strand)

Markers analyzed: 28

Coverage: A total of 28 specific disease, pharmacogenomic, nutrigenomic, and trait markers were evaluated; call rate reported as 98.6%. ADH1B rs1229984 resulted in a 'No call'.

Limitations: Genome-wide SNP array genotyping interrogates predefined variants and tag SNPs; it does not constitute clinical diagnostic sequencing, does not capture structural or copy number variants (such as CYP2D6 duplications/deletions), and may miss rare pathogenic variants.

Findings by area

Cardiovascular and Lipid Risk Architecture Elevated

The patient carries several synergistic risk alleles impacting atherogenic lipid transport, vascular calcification, and myocardial infarction risk, warranting formal baseline lipid profiling and circulating Lp(a) quantification.

Gene	rsID	Genotype	Phenotype	Risk	Evidence	Explanation / Recommendation
APOE	rs429358	CT	APOE e3/e4 genotype	Elevated	Established	Heterozygosity for the e4-defining allele (alongside rs7412 CC) establishes an e3/e4 phenotype, associated with higher circulating LDL cholesterol, heightened cardiovascular disease risk, and accelerated atherogenesis. *Obtain fasting lipid panel; consider tighter LDL-C targets and monitor cardiovascular risk factors proactively.*
APOE	rs7412	CC	e2 allele not detected	Typical	Established	Absence of the protective e2 allele confirms standard clearance baseline relative to the protective allele. *Incorporate within overall APOE e3/e4 risk stratification.*
LPA	rs10455872	AG	Elevated lipoprotein(a) risk allele	Elevated	Established	Carriage of the minor G allele at rs10455872 is strongly linked to elevated circulating levels of lipoprotein(a) and heightened independent atherosclerotic cardiovascular risk. *Measure serum lipoprotein(a) once to establish lifetime baseline risk.*
CDKN2B-AS1	rs1333049	CG	9p21.3 coronary artery disease susceptibility	Slightly Elevated	Established	Heterozygosity at 9p21.3 is an established independent genetic marker conferring modest incremental susceptibility to coronary artery disease. *Factor into global atherosclerotic cardiovascular disease (ASCVD) risk estimation.*

Pharmacogenomics and Drug Metabolism High

Clinically actionable variants were identified across several major drug-metabolizing enzymes and transporters, including CYP2C19, CYP2C9, VKORC1, and SLCO1B1.

Gene	rsID	Genotype	Phenotype	Risk	Evidence	Explanation / Recommendation

SLCO1B1	rs4149056	CT	*1/*5 - decreased function	High	Established	The *5 allele impairs OATP1B1 hepatic uptake transporter function, significantly increasing systemic statin exposure, particularly for simvastatin, and elevating risk of statin-induced myopathy or rhabdomyolysis. *Consult CPIC guidelines regarding statin selection and dosing if lipid-lowering therapy is indicated; consider alternative statins (e.g., rosuvastatin or pravastatin) or non-statin therapies.*
CYP2C19	rs4244285	AG	*1/*2 - intermediate metabolizer	Elevated	Established	Carriage of the loss-of-function *2 allele reduces bioactivation of prodrugs such as clopidogrel and slows clearance of certain proton pump inhibitors and select SSRIs. *Follow CPIC guidelines for antiplatelet therapy selection; alternative agents such as prasugrel or ticagrelor may be indicated if antiplatelet therapy is required.*
CYP2C19	rs12248560	CC	*17 allele not detected	Typical	Established	Absence of the increased-function *17 allele confirms that CYP2C19 phenotype is governed by the *2 allele. *Classify CYP2C19 as intermediate metabolizer overall.*
CYP2C9	rs1799853	CT	*1/*2 - intermediate metabolizer	Elevated	Established	The CYP2C9*2 allele reduces catalytic clearance of CYP2C9 substrates, including warfarin, phenytoin, and certain NSAIDs. *Anticipate lower maintenance requirements for sensitive CYP2C9 substrates.*
CYP2C9	rs1057910	AA	*3 allele not detected	Typical	Established	Absence of *3 allele confirms lack of additional CYP2C9 clearance reduction from this locus. *Maintain *1/*2 classification.*

VKORC1	rs9923231	CT	-1639G>A intermediate warfarin sensitivity	Elevated	Established	The A allele downregulates VKORC1 expression, increasing sensitivity to vitamin K antagonists. *Utilize CPIC pharmacogenetic warfarin dosing algorithms combining CYP2C9 and VKORC1 genotypes if vitamin K antagonist therapy is contemplated.*
CYP1A2	rs762551	AC	*1F intermediate/slower caffeine metabolizer	Informational	Probable	Heterozygosity for the *1F variant predicts intermediate CYP1A2 inducibility and moderate rate of caffeine clearance. *Advise moderation in caffeine intake if experiencing sleep disturbances or palpitations.*
ABCG2	rs2231142	GG	Normal ABCG2 transporter function	Typical	Established	The Q141K variant was not detected, predicting normal BCRP transporter activity. *Standard dosing expectations for BCRP substrates.*
G6PD	rs1050828	CC	G6PD A- not detected	Typical	Established	The common African-origin deficiency allele A- (V68M) was not detected. *Standard clinical surveillance for hemolytic triggers.*
ADH1B	rs1229984	--	No call	Informational	Preliminary	Marker could not be genotyped by the array platform. *No clinical inference should be made from this locus.*
ALDH2	rs671	GG	Normal ALDH2 activity (*1/*1)	Typical	Established	The *2 deficiency allele was not detected; normal acetaldehyde oxidation capacity. *Standard clinical guidance.*

Carrier Status and Monogenic Disease Markers Slightly Elevated

Identified as a heterozygous carrier for HFE-associated hereditary hemochromatosis. Common thrombophilic and cystic fibrosis mutations were not detected.

Gene	rsID	Genotype	Phenotype	Risk	Evidence	Explanation / Recommendation

HFE	rs1800562	AG	C282Y carrier	Slightly Elevated	Established	Heterozygosity for HFE C282Y confers carrier status for autosomal recessive hereditary hemochromatosis. Simple carriers rarely develop significant iron overload without comorbid risk factors. *Confirm with validated clinical diagnostic genetic testing; evaluate baseline fasting transferrin saturation and serum ferritin.*
HFE	rs1799945	CC	H63D not detected	Typical	Established	The absence of H63D rules out compound heterozygosity (C282Y/H63D). *Standard monitoring.*
F5	rs6025	CC	Factor V Leiden not detected	Typical	Established	Negative for the Factor V Leiden thrombophilia variant. *Standard clinical assessment of venous thromboembolism risk.*
F2	rs1799963	GG	Prothrombin G20210A not detected	Typical	Established	Negative for the prothrombin 20210A thrombophilia variant. *Standard clinical assessment.*
CFTR	rs113993960	II	F508del not detected	Typical	Established	Common cystic fibrosis delta F508 deletion was not detected. *Routine clinical oversight.*

Nutrigenomics and Food Response Slightly Elevated

Genotype indicates lactase non-persistence, non-secretor status for mucosal histo-blood antigens, mildly reduced MTHFR activity, and altered vitamin D carrier protein binding.

Gene	rsID	Genotype	Phenotype	Risk	Evidence	Explanation / Recommendation
MCM6/LCT	rs4988235	GG	Lactase non-persistence	Informational	Established	The GG genotype at -13910 is characteristic of adult lactase non-persistence, predisposing to primary lactose intolerance. *Guide dietary lactose restriction or lactase enzyme supplementation according to gastrointestinal symptoms; ensure adequate calcium and vitamin D intake.*

MTHFR	rs1801133	AG	C677T heterozygous	Slightly Elevated	Established	Heterozygosity for C677T confers approximately 30-35% reduced MTHFR enzymatic activity, which may mildly affect remethylation of homocysteine to methionine under conditions of folate insufficiency. *Assess serum folate, vitamin B12, and plasma total homocysteine; prioritize natural dietary folate sources.*
MTHFR	rs1801131	TT	A1298C not detected	Typical	Established	Absence of the A1298C variant eliminates compound heterozygosity at this locus. *Standard nutritional monitoring.*
GC	rs2282679	GT	Altered vitamin D binding protein affinity	Informational	Probable	The T allele is associated with lower circulating 25-hydroxyvitamin D concentrations due to altered vitamin D binding protein levels. *Monitor circulating 25-hydroxyvitamin D levels periodically.*
FADS1	rs174546	CT	Intermediate fatty acid desaturase conversion	Informational	Probable	Heterozygous genotype is associated with moderate endogenous conversion of plant alpha-linolenic acid into active long-chain polyunsaturated fatty acids (EPA/DHA). *Consider preformed dietary sources of omega-3 fatty acids (EPA/DHA).*
FUT2	rs601338	AA	Non-secretor status (W143X homozygote)	Informational	Established	Homozygosity for the nonsense variant results in absence of ABO blood group antigens in bodily fluids and mucosa, influencing gut microbiome diversity and conferring resistance to norovirus GI/GII strains. *Support intestinal barrier integrity and diverse dietary prebiotic intake.*

Metabolic and Endocrine Profile Slightly Elevated

Polygenic susceptibility markers show modest predisposition to type 2 diabetes and adiposity, relevant in the context of general metabolic oversight and thyroid tracking.

Gene	rsID	Genotype	Phenotype	Risk	Evidence	Explanation / Recommendation

TCF7L2	rs7903146	CT	Increased type 2 diabetes risk allele	Slightly Elevated	Established	Heterozygosity for the risk allele at TCF7L2 is one of the strongest common genetic predictors of impaired beta-cell function and type 2 diabetes. *Periodically assess fasting blood glucose and HbA1c.*
FTO	rs9939609	AT	Elevated adiposity predisposition	Slightly Elevated	Established	The A allele is associated with altered satiety regulation and modest polygenic risk of elevated BMI. *Emphasize dietary satiety strategies and regular physical activity.*
PPARG	rs1801282	CC	Pro12Pro common allele	Typical	Established	Carries the wild-type Pro allele without the protective insulin-sensitizing Ala variant. *Standard lifestyle and insulin sensitivity maintenance.*
PNPLA3	rs738409	CG	I148M heterozygous	Slightly Elevated	Established	Heterozygous carrier of the G allele, associated with reduced hepatic triglyceride hydrolysis and mildly increased susceptibility to hepatic steatosis. *Monitor hepatic enzymes (ALT, AST) periodically; limit refined fructose and alcohol consumption.*

Cellular Defense, Neurochemistry, and Fitness Traits Informational

Heterozygous profiles across antioxidant and catecholamine pathways with mixed muscle performance potential.

Gene	rsID	Genotype	Phenotype	Risk	Evidence	Explanation / Recommendation
COMT	rs4680	AG	Val158Met intermediate activity	Informational	Established	Intermediate enzymatic clearance of catecholamines (dopamine, epinephrine) and catechol estrogens. *Support balanced stress management and routine sleep hygiene.*
SOD2	rs4880	AG	Ala16Val intermediate mitochondrial targeting	Informational	Probable	Heterozygosity results in intermediate mitochondrial superoxide dismutase transport. *Promote diet rich in dietary antioxidants and polyphenols.*
GSTP1	rs1695	AG	Ile105Val intermediate GST activity	Informational	Probable	Intermediate phase II glutathione conjugation capacity. *Encourage cruciferous vegetable consumption.*

ACTN3	rs1815739	CT	R577X heterozygote	Informational	Established	Presence of one functioning R allele maintains alpha-actinin-3 expression in fast-twitch muscle fibers, supporting a balanced endurance and power athletic profile. *Suitable for mixed aerobic and resistance conditioning.*
TNF	rs1800629	GG	-308G baseline expression	Typical	Established	Wild-type genotype; lacks the hyper-inflammatory -308A promoter allele. *Standard clinical surveillance.*
HLA-DQA1	rs2187668	CC	DQ2.5 tag not detected	Typical	Established	Negative for the primary HLA-DQ2.5 risk tag associated with celiac disease, indicating high negative predictive value. *Routine clinical oversight.*
HERC2	rs12913832	AG	Intermediate iris pigmentation marker	Informational	Established	Heterozygous genotype at the primary regulatory region for OCA2 expression, commonly associated with green, hazel, or intermediate eye color. *None; physical trait.*

Disease risks

Atherosclerotic Cardiovascular Disease (ASCVD) Elevated

Genes: APOE, LPA, CDKN2B-AS1

Synergy between APOE e4, LPA rs10455872, and 9p21.3 risk alleles elevates baseline lifetime risk for atherogenesis and coronary events.

Actions:

- Quantify serum lipoprotein(a) and comprehensive fasting lipid profile.
- Maintain strict control of blood pressure, glycemic parameters, and lifestyle factors.

Late-Onset Alzheimer's Disease Slightly Elevated

Genes: APOE

The APOE e3/e4 genotype confers an approximately 3-fold higher lifetime risk of late-onset Alzheimer's disease compared to the baseline e3/e3 genotype, though it is not deterministic.

Actions:

- Prioritize vascular and metabolic risk factor control, regular aerobic exercise, and cognitive engagement.
- Ensure adequate sleep hygiene.

Type 2 Diabetes Mellitus Slightly Elevated

Genes: TCF7L2, FTO

Carriage of risk alleles at TCF7L2 and FTO contributes to polygenic vulnerability for impaired insulin secretion and metabolic dysregulation.

Actions:

- Incorporate annual fasting glucose or HbA1c screening into primary care follow-up.

Hereditary Hemochromatosis Slightly Elevated

Genes: HFE

Heterozygous carrier for HFE C282Y. Clinical penetrance is very low in single heterozygotes, but iron accumulation can occur in the presence of secondary modifiers.

Actions:

- Obtain baseline fasting transferrin saturation and serum ferritin.
- Recommend clinical diagnostic confirmation before initiating specific interventions.

Carrier status

Condition	Gene	rsID	Status	Note
Hereditary Hemochromatosis (HFE-related)	HFE	rs1800562	Carrier	Identified as heterozygous for C282Y (rs1800562 AG). Confirmatory testing via an accredited clinical diagnostic method is required. Genetic counseling is advised for family planning.
Cystic Fibrosis (CFTR-related)	CFTR	rs113993960	Not Detected	Common delta F508 variant was not detected; this does not rule out rare pathogenic CFTR variants not evaluated by this array.
Factor V Leiden Thrombophilia	F5	rs6025	Not Detected	Negative for Factor V Leiden (R506Q).
Prothrombin G20210A Thrombophilia	F2	rs1799963	Not Detected	Negative for prothrombin 20210A mutation.

Pharmacogenomics

Gene	rsID	Predicted phenotype	Affected drugs	Recommendation
SLCO1B1	rs4149056	SLCO1B1 *1/*5 (Decreased function)	Simvastatin, Atorvastatin, Pravastatin, Rosuvastatin	Marked increase in simvastatin plasma concentrations and myopathy risk. Per CPIC guidelines, consider an alternative statin (e.g., rosuvastatin or pravastatin at recommended doses) if lipid therapy is indicated, or limit simvastatin dose.
CYP2C19	rs4244285, rs12248560	CYP2C19 *1/*2 (Intermediate metabolizer)	Clopidogrel, Omeprazole, Esomeprazole, Citalopram, Sertraline	Suboptimal activation of clopidogrel with diminished antiplatelet effect; consider alternative P2Y12 inhibitors (e.g., ticagrelor, prasugrel) per CPIC guidelines if indicated. Standard or adjusted dosing may be needed for specific PPIs and SSRIs.

CYP2C9	rs1799853, rs1057910	CYP2C9 *1/*2 (Intermediate metabolizer)	Warfarin, Phenytoin, Celecoxib, Ibuprofen	Reduced clearance of CYP2C9 substrates. In combination with VKORC1 genotype, lower initial doses of warfarin are anticipated if anticoagulant therapy is needed.
VKORC1	rs9923231	-1639G>A (Intermediate warfarin sensitivity)	Warfarin	Increased sensitivity to vitamin K antagonists; use genotype-guided pharmacogenetic warfarin dosing tools to guide initial therapeutic titration.
CYP1A2	rs762551	CYP1A2 *1F (Intermediate / slower caffeine inducer)	Caffeine, Theophylline, Clozapine, Olanzapine	Moderate metabolic clearance rate for CYP1A2 substrates; monitor for sensitivity to high caffeine intake.

Nutrigenomics

Nutrient	Genes	Finding	Recommendation
Lactose	MCM6/LCT	rs4988235 GG genotype indicates genetically programmed adult lactase non-persistence.	Consider trial of lactose restriction or lactase enzyme supplementation if GI symptoms occur; ensure adequate dietary calcium.
Folate	MTHFR	rs1801133 AG (C677T heterozygous) confers modest reduction in homocysteine remethylation efficiency.	Prioritize folate-dense leafy green vegetables and whole foods; assess homocysteine and serum folate status before considering dietary supplements.
Vitamin D	GC	rs2282679 GT indicates potential for lower circulating 25-hydroxyvitamin D concentrations via altered binding protein affinity.	Measure serum 25-hydroxyvitamin D to establish sufficiency before considering supplementation.
Omega-3 Fatty Acids	FADS1	rs174546 CT is associated with intermediate endogenous desaturation of alpha-linolenic acid to EPA and DHA.	Encourage consumption of dietary long-chain polyunsaturated fats from marine sources.

Traits

Trait	Gene	Finding
Secretor Status	FUT2	rs601338 AA non-secretor genotype; lacks ABH histo-blood antigens in mucosal secretions, conferring relative norovirus resistance and distinct gut microbiota composition.
Muscle Fiber Performance	ACTN3	rs1815739 CT heterozygous (R577X); produces alpha-actinin-3, conferring mixed capacity for power, sprint, and endurance activities.
Catecholamine Metabolism	COMT	rs4680 AG (Val158Met); confers intermediate enzymatic breakdown rate for catecholamines and estrogens.
Eye Color	HERC2	rs12913832 AG genotype; associated with intermediate or variable iris pigmentation.

Lifestyle recommendations

- Adopt an anti-atherogenic, heart-healthy dietary pattern (e.g., Mediterranean diet) rich in unprocessed foods, unsaturated fats, and soluble fiber to mitigate APOE e4 and LPA-related cardiovascular risks.
- Engage in structured aerobic conditioning (minimum 150 minutes of moderate-intensity activity weekly) combined with resistance training to optimize insulin sensitivity and metabolic health.
- Moderate dietary dairy intake or utilize lactase enzyme supplements if abdominal discomfort, bloating, or diarrhea occur following dairy consumption.
- Avoid unprescribed iron supplementation and unnecessary vitamin C megadoses given the presence of the HFE

- Avoid unprescribed iron supplementation and unnecessary vitamin C megadoses given the presence of the HFE C282Y carrier allele.
- Moderate daily caffeine consumption if prone to sleep latency prolongation or tachyarrhythmias.

Recommended tests

Test	Reason
Lipoprotein(a) [Lp(a)]	Indicated by LPA rs10455872 heterozygous genotype to evaluate independent cardiovascular risk.
Comprehensive Lipid Panel (TC, HDL-C, LDL-C, Triglycerides, Non-HDL)	Indicated due to APOE e3/e4 genotype and CDKN2B-AS1 risk alleles.
Iron Studies (Serum Iron, TIBC, Transferrin Saturation, Serum Ferritin)	Indicated to evaluate baseline iron balance following the detection of the HFE C282Y carrier state.
Fasting Plasma Glucose and HbA1c	Indicated for metabolic baseline assessment in view of the TCF7L2 risk variant.
Serum 25-Hydroxyvitamin D, Total Homocysteine, and Folate	Indicated to assess functional micronutrient status given GC and MTHFR C677T findings.
Thyroid Function Tests (TSH, Free T4, +/- Free T3)	Indicated in accordance with the established clinical context of ongoing thyroid monitoring.
Clinical Diagnostic Confirmatory Genotyping	Mandatory diagnostic confirmation of HFE C282Y and key pharmacogenetic variants (CYP2C19, SLCO1B1) before definitive clinical decisions.

Red flags

- Severe, unexplained muscle pain, weakness, or dark-colored urine if exposed to statin therapy (particularly simvastatin), necessitating immediate clinical evaluation for myopathy/rhabdomyolysis.
- Recurrent signs of systemic iron overload (unexplained fatigue, arthralgias, skin bronzing, elevated transaminases) warranting prompt serum ferritin and transferrin saturation testing.
- Discrepancy noted between clinician profile (43-year-old male) and test requisition metadata (female, born 1984); identity and sample provenance must be verified prior to clinical action.

Limitations

Array genotyping provides targeted analysis of specific single-nucleotide polymorphisms and does not sequence fu genes, identify copy number alterations, structural variants, or detect rare private mutations. Genotypic risk estimations are probabilistic and modified by environment, age, sex, and lifestyle. Any carrier or pharmacogenomic finding must be clinically verified via CLIA/CAP-accredited diagnostic molecular testing before modifying patient care.