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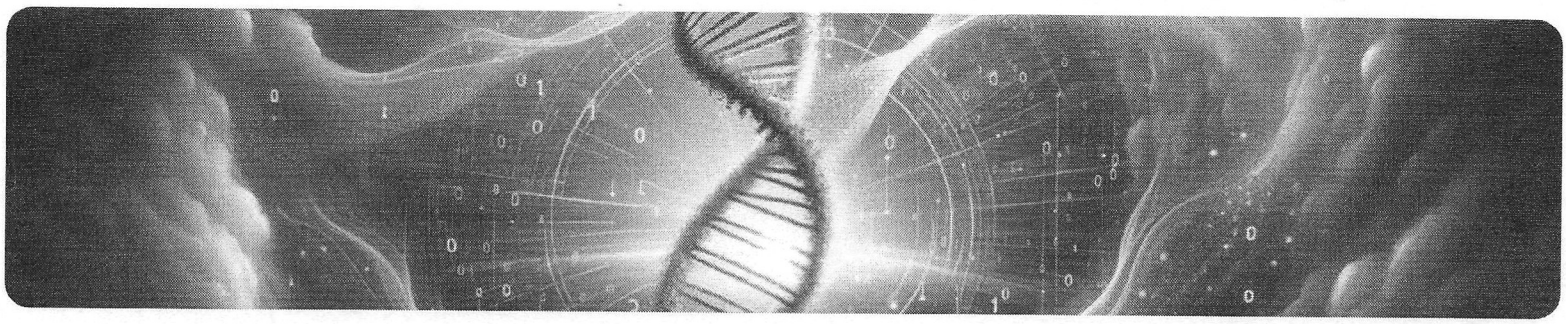
Next-Gen Disease Screen

Summary Report BETA



Genome: **Michael Vaughn**
Report Date: **August 17, 2026**
Analysis: **Next-Gen Disease Screen Premium**
Data: **Whole Genome Sequencing**

Confidential Information

Genome Sequencing Laboratory Information

- CLIA Certification: **45D1102202**
- CAP Accreditation: **7212851**
- Location: **USA** (Houston, Texas)
- Service: **30× Whole Genome Sequencing**

Genome Data Analysis Information

- Analysis Performed By: **Sequencing.com**
- Data Analyzed: **30× Whole Genome Sequencing**
- Email: **support@sequencing.com**
- Phone: **1-833-544-0001**

Simvastatin Response – Toxicity

Gene: SLCO1B1
Variant Identifier: rs4149056, RCV001787325 Gene Variant rs4149056
Your Data: TC
Risk Status: Medication Carrier or Detected

Description: Simvastatin response - Toxicity, commonly known as statin toxicity, refers to a negative reaction to simvastatin, a medication used to lower cholesterol levels and reduce the risk of heart disease. Statin toxicity can cause muscle and liver damage, usually due to high doses or interactions with other medications. Treatment options may include reducing the dosage, switching to a different statin, or using alternative cholesterol-lowering medications. Regular monitoring of liver and muscle enzymes, as well as discussing any concerns with a healthcare provider, can help manage and prevent simvastatin toxicity.

Symptoms: muscle pain or weakness, dark-colored urine, unexplained muscle tenderness or weakness, fever, unusual tiredness, abdominal pain, nausea, vomiting, loss of appetite, yellowing of the skin or eyes (jaundice), dark urine, pale stools, confusion, mèmory problems, difficulty breathing, chest pain, swelling in the ankles or feet

HMG CoA Reductase Inhibitors Response – Toxicity

Gene: SLCO1B1
Variant Identifier: rs4149056, RCV001787333
Your Data: TC
Risk Status: Medication Carrier or Detected

Description: HMG-CoA reductase inhibitors, commonly known as statins, are medications used to lower cholesterol levels in the blood, but some individuals may experience toxicity as a response to these drugs. This condition, often referred to as statin intolerance or statin-associated muscle symptoms (SAMS), occurs when the body reacts negatively to the medication, potentially leading to complications. Treatment options for managing statin toxicity include adjusting the dosage, switching to a different statin, or trying alternative cholesterol-lowering medications such as ezetimibe or PCSK9 inhibitors. In some cases, lifestyle changes such as diet and exercise may also be recommended to help manage cholesterol levels without the need for statins.

Gene: *SLCO1B1*

Go to:

The *SLCO1B1* gene, located on the short arm of chromosome 12, encodes OATP1B1, one of the major hepatic influx transport proteins. Primarily expressed on the basolateral surface of hepatocytes, OATP1B1 facilitates the uptake of various endogenous and exogenous compounds including bile acids, thyroid hormones, bilirubin, methotrexate, angiotensin-converting enzyme inhibitors, angiotensin II receptor blockers, and statins. Decreased expression or function of OATP1B1 can lead to increased plasma concentrations of these substrates, potentially resulting in systemic drug toxicity or adverse effects. Genetic variations at the *SLCO1B1* locus, as well as drug–drug interactions, can change OATP1B1 function and increase an individual's risk for adverse effects from these substrate medications.

***Sequencing.com identified a genetic variant in the SLCO1B1 gene (rs4149056) aka (*5 variant).**

Nomenclature for Selected *SLCO1B1* Alleles

Go to:

Common allele name	Alternative names	HGVS reference sequence		dbSNP reference identifier for allele location
		Coding	Protein	
*SLCO1B1*1*	SLCO1B1*1A	NM_006446.5	NP_006437.3	
*SLCO1B1*4*	35305C>A (P155T)	NM_006446.5:c.463C>A	NP_006437.3:p.Pro155Thr	rs11045819
*SLCO1B1*5*	37041T>C (V174A)	NM_006446.5:c.521T>C	NP_006437.3:p.Val174Ala	rs4149056
*SLCO1B1*9*	64425G>C (G488A)	NM_006446.5:c.1463G>C	NP_006437.3:p.Gly488Ala	rs59502379
*SLCO1B1*14*[a]	35230A>G (N130D)	NM_006446.5:c.388A>G	NP_006437.3:p.Asn130Asp	rs2306283
	35305C>A (P155T)	NM_006446.5:c.463C>A	NP_006437.3:p.Pro155Thr	rs11045819
*SLCO1B1*15*[b]	35230A>G (N130D)	NM_006446.5:c.388A>G	NP_006437.3:p.Asn130Asp	rs2306283
	37041T>C (V174A)	NM_006446.5:c.521T>C	NP_006437.3:p.Val174Ala	rs4149056
*SLCO1B1*20*[c]	35230A>G (N130D)	NM_006446.5:c.388A>G	NP_006437.3:p.Asn130Asp	rs2306283
	97468A>C (L643F)	NM_006446.5:c.1929A>C	NP_006437.3:p.Leu643Phe	rs34671512
*SLCO1B1*37*	SLCO1B1-*1B, *1F, *1G, *1H 35230A>G (N130D)	NM_006446.5:c.388A>G	NP_006437.3:p.Asn130Asp	rs2306283

Each person has 2 gene alleles, one derived from each parent. The *5 allele variant is an allele that produces no functional protein. It is paired with a normal functioning allele *1 and so this transporter protein level is expressed in the body at about 50% of normal levels.

Functional Classification of *SLCO1B1* Allele (CPIC, 2021)

Allele clinical functional status	***SLCO1B1* star alleles**
Increased function	*14, *20
Normal function	*1, *37
No function	*5, *9, *15, *23, *31, *46, *47, *48, *49
Unknown function	*43, *44, *45

Selected *SLCO1B1* Phenotype-Genotype Predictions (CPIC, 2022)

Clinical phenotype	**Example *SLCO1B1* genotype**	**Clinical alert category[a]**
Normal function	*1/*1 *1/*14 *1/*37	Low risk
Increased function	*14/*14 *14/*20 *20/*20	Low risk
Decreased function	*1/*5 *1/*9 *15/*37	High risk
Possible decreased function	*2/*5 *2/*15 *3/*9	High risk
Poor function	*5/*5 *5/*9 *9/*23 *15/*15	High risk

Consequences:

This protein (SLCO1B1) transports certain medications out of the blood stream and into liver cells (hepatocytes) where the drug are metabolized (broken-down). If medications such thyroid hormones, ACE-inhibitors and ARB blood pressure medications and Statins are taken, blood levels will build-up. If these medications are taken at normal doses, toxic effects from high blood levels are likely to occur. Even at low doses, these medications may cause exaggerated effects in persons with the identified genotype *1/*5. This is particularly dangerous when "Statins" (used for the treatment of high cholesterol) are taken as high levels can cause muscle damage "statin-associated muscle symptoms" (SAMS).