MAYO CLINIC

Mayo Clinic GeneGuide™

Report

Charles Warden
DOB: **04/05/1985**

Table Of Contents

Demographics

Consumer ID
B48A4583-F87E-4871-895F-B9ED7AB76B2B

Gender
Male

Consumer Name
Charles Warden

Age at Time of Collection
33 years

Consumer Phone
404-316-0012

Consumer Email
cwarden45@gmail.com

Date of Birth
04/05/1985

Order Information

Helix Sample ID
1645-20190128150501

Specimen Description
Saliva

Requested by
Ramos, Carlos

Date Service Requested
28 Jan 2019

Specimen Received Date
28 Jan 2019

Specimen Collected Date
28 Jan 2019

Lab Director:
Matthew J. Ferber, Ph.D. FACMGG

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Reading and Interpreting this Genetic Test Result Report

GeneGuide Genetic Test Result printable reports are designed to help you easily communicate your results with your healthcare provider or anyone else you may want to share your report with. GeneGuide Genetic Test Result reports are separated into the following categories:

- Carrier Screening
- Medication Response
- Disease Risk
- Health Traits

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Introduction to Carrier Screening

Carrier screening is a type of genetic testing that lets you know if you carry a gene variant you could pass along to your children, possibly affecting their health, depending upon the genes inherited from their other parent. This type of testing can be useful if you're planning a family. All people have a chance of being carriers for genetic conditions. People in some ethnic groups have increased risk of specific genetic conditions, but all individuals may benefit from carrier screening before starting a family.

In most cases, being a carrier for one of the conditions in the Carrier Screening category in Mayo Clinic GeneGuide™ does not affect your health. That's because the variants identified in carrier screening cause health problems only in people who inherit a variant in the same gene from both parents. This pattern is known as autosomal recessive inheritance.

If you carry a variant in this category and your partner does not, each of your children has a 1 in 2 (50%) chance of inheriting the variant and being a carrier as well. A child in this scenario, however, has no chance of being affected by the associated genetic condition.

On the other hand, if both parents carry a variant in this gene, each child has a 1 in 4 (25%) chance of inheriting a copy of the variant from both parents. If this happens, the child has the condition. If you are a carrier for a genetic condition in the Carrier Screening category, your partner may want to get tested. If you are both carriers, you can use that knowledge to help you prepare for pregnancy and make choices that are right for you and your family.

Carrier Screening Result Summary

Condition	Gene	# Variants	Variant(s) Identified	Variant(s) Classification	Interpretation
Cystic Fibrosis	CFTR	0			No variants identified
GJB2-related hearing loss	GJB2	0			No variants identified
MCAD Deficiency	ACADM	0			No variants identified
Sickle Cell Disease	HBB	0			No variants identified

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Cystic Fibrosis Results

Condition	Gene	# Variants	Variant(s) Identified	Variant(s) Classification	Interpretation
Cystic Fibrosis	CFTR	0			No variants identified

Cystic Fibrosis interpretation

We ruled out the most common genetic variants in the CFTR gene. Even though this test didn't identify any variants, there is a small chance that you still could be a carrier. The risk level depends in part on your ethnic background, as noted below in the residual risk information. For most people, there's nothing more you need to do based on this result. If you have a family history of cystic fibrosis, you may want to consider more comprehensive testing. Talk with your health care provider for information about comprehensive testing for cystic fibrosis.

Cystic fibrosis is a genetic condition that primarily affects the respiratory (lungs), digestive (stomach, intestines, pancreas) and reproductive systems (sex organs). Cystic fibrosis is an inherited genetic disease caused by variants in the CFTR gene located on chromosome 7. Most individuals with cystic fibrosis have two variants in the CFTR gene.

This test is not intended to diagnose a disease or tell you anything about your risk for developing a disease in the future. A genetic consultation may be of benefit. Please contact the independent genetic counseling provider who reviewed your test order and results, PWNHealth toll free at 1-888-494-7333 or by email at gc@pwnhealth.com, if you have questions regarding this report.

Residual risk information

Chance of having a genetic variant based on this test:
- Non-Hispanic White: 1 in 200 (0.5%)
- Ashkenazi Jewish: 1 in 380 (0.3%)
- Hispanic White: 1 in 200 (0.5%)
- Asian-American: 1 in 180 (0.6%)
- African-American: 1 in 170 (0.6%)
- Other/Mixed race: No specific data is available.

Method: The variants interpreted by the Mayo Clinic GeneGuide™ system are detected by our partner, Helix. The Helix Exome+ assay uses next generation sequencing (NGS) of DNA to analyze the entire human exome plus several hundred thousand additional informative regions outside of the exome, including the complete mitochondrial genome. The Helix Exome+ assay was performed on genomic DNA extracted from your submitted saliva sample. DNA from your sample was captured and enriched using a custom set of reagents for target selection. Next Generation Sequencing was performed on an Illumina DNA sequencing instrument using HapMap samples NA12878 and NA12877 as internal controls. Alignment to a modified version of GRCh38 and variant calling were completed using a customized version of Sentieon's DNAseq software. The Helix bioinformatics pipeline applied several quality control filters to produce the final passing variants list. Using a semi-automated platform for laboratory review and reporting, The Mayo Clinic GeneGuide™ interpretive system then pulled a predetermined set of clinically validated variants from the Helix Secure Database, and matched the variants to the appropriate result interpretation.

Variants Tested: The variant nomenclature that is used to describe the Cystic Fibrosis variants included in Mayo Clinic GeneGuide™ is based on:

NM_000492.3(CFTR)

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

c.254G>A (p.Gly85Glu), c.350G>A (p.Arg117His), c.489+1G>T, c.579+1G>T, c.1000C>T (p.Arg334Trp), c.1040G>C (p.Arg347Pro), c.1364C>A (p.Ala455Glu), c.1519_1521delATC (p.Ile507del), c.1521_1523delCTT (p.Phe508del), c.1521_1523delCTT (p.Phe508del), c.1521_1523delCTT (p.Phe508del), c.1585-1G>A, c.1624G>T (p.Gly542Ter), c.1652G>A (p.Gly551Asp), c.1657C>T (p.Arg553Ter), c.1679G>C (p.Arg560Thr), c.1766+1G>A, c.2052delA (p.Lys684Asnfs), c.2657+5G>A, c.2988+1G>A, c.3484C>T (p.Arg1162Ter), c.3528delC (p.Lys1177Serfs), c.3717+12191C>T, c.3846G>A (p.Trp1282Ter), c.3909C>G (p.Asn1303Lys)

Cautions:

- The interpretation of this report is based upon the information provided by the ordering physician at the time of testing. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.
- This test is designed to detect the specific clinically validated variants listed in this report. It is possible that other types of disease-causing variants exist in this or other related genes that were not detected.
- Patients who have ever had an allogeneic blood or marrow transplant or who have received a heterologous blood transfusion within the last month, can have inaccurate genetic test results due to presence of donor DNA. Patients who have undergone liver transplantation may also have inaccurate genetic test results.
- Variant nomenclature is based on genomic build GRCh38. Variants are evaluated and classified according to ACMG recommendations (Richards, et al., 2015). Variant classification may change over time. If a variant that was part of Mayo Clinic GeneGuide™ is reclassified and you have questions, please contact us at 1-877-858-0398.

Cautions for Health Care Providers:

- This test is not intended to diagnose a disease, determine medical treatment, or tell the user anything about their current state of health.
- This test is intended to provide users with their genetic information to inform lifestyle decisions and conversations with their doctor or other healthcare professional.
- Any diagnostic or treatment decisions should be based on testing and/or other information that your healthcare provider determines to be appropriate for you.

Warnings:

- The test is intended only for autosomal recessive carrier screening in adults of reproductive age.
- The test does not detect all genetic variants related to the condition, and the absence of a variant tested does not rule out the presence of other genetic variants in this, or other genes, that may be disease related
- The results of this test are intended to be interpreted by a board-certified clinical molecular geneticist or equivalent and should be used in conjunction with other available laboratory and clinical information.
- This test is not intended for disease diagnosis, prenatal testing of fetuses, risk assessment, prognosis or pre-symptomatic testing, susceptibility testing, or newborn screening.
- This test is not intended to diagnose a disease, or tell you anything about your risk for developing a disease in the future. On its own, this test is also not intended to tell you anything about the health of your fetus, or your newborn child’s risk of developing a particular disease later on in life.
- This test is not a substitute for visits to a healthcare provider. It is recommended that you consult with a healthcare provider if you have any questions or concerns about your results.
- The test does not diagnose any health conditions. Results should be used along with other clinical information for any medical purposes
- Your ethnicity may affect how your genetic health results are interpreted

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

GJB2-related hearing loss Results

Condition	Gene	# Variants	Variant(s) Identified	Variant(s) Classification	Interpretation
GJB2-related hearing loss	GJB2	0			No variants identified

GJB2-related hearing loss interpretation

We ruled out the most common genetic variants in the GJB2 gene. Even though this test didn't identify any variants, there is a small chance that you still could be a carrier. The risk level depends in part on your ethnic background, as noted below in the residual risk information. For most people, there's nothing more you need to do based on this result. If you have a family history of inherited hearing loss related to GJB2 variants, you may want to consider more comprehensive testing. Talk with your health care provider for information about comprehensive testing for GJB2 variants.

Variants in many different genes are associated with inherited hearing loss. In Mayo Clinic GeneGuide™, we test for inherited nonsyndromic hearing loss caused by variants in the GJB2 gene on chromosome 13. GJB2 variants are the most common cause of autosomal recessive nonsyndromic hearing loss worldwide. GJB2-related hearing loss is typically present at birth, is slowly progressive, and can be mild to severe. Most people with inherited hearing loss related to GJB2 variants have two associated variants.

This test is not intended to diagnose a disease or tell you anything about your risk for developing a disease in the future. A genetic consultation may be of benefit. Please contact the independent genetic counseling provider who reviewed your test order and results, PWNHealth toll free at 1-888-494-7333 or by email at gc@pwnhealth.com, if you have questions regarding this report.

Residual Risk Information

Chance of having a genetic variant based on this test:
- Non-Hispanic White: Less than 1 in 160 (0.6%)
- Ashkenazi Jewish: Data are not available on how much risk is reduced.
- Hispanic White: Data are not available on how much risk is reduced.
- Asian-American: Data are not available on how much risk is reduced.
- African-American: Data are not available on how much risk is reduced.
- Other/Mixed race: Data are not available on how much risk is reduced.

Method: The variants interpreted by the Mayo Clinic GeneGuide™ system are detected by our partner, Helix. The Helix Exome+ assay uses next generation sequencing (NGS) of DNA to analyze the entire human exome plus several hundred thousand additional informative regions outside of the exome, including the complete mitochondrial genome. The Helix Exome+ assay was performed on genomic DNA extracted from your submitted saliva sample. DNA from your sample was captured and enriched using a custom set of reagents for target selection. Next Generation Sequencing was performed on an Illumina DNA sequencing instrument using HapMap samples NA12878 and NA12877 as internal controls. Alignment to a modified version of GRCh38 and variant calling were completed using a customized version of Sentieon's DNAseq software. The Helix bioinformatics pipeline applied several quality control filters to produce the final passing variants list. Using a semi-automated platform for laboratory review and reporting, The Mayo Clinic GeneGuide™ interpretive system then pulled a predetermined set of clinically validated variants from the Helix Secure Database, and matched the variants to the appropriate result interpretation.

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Variants Tested: The variant nomenclature that is used to describe the GJB2-related hearing loss variants included in Mayo Clinic GeneGuide™ is based on:

NM_004004.5(GJB2)

c.358_360delGAG (p.Glu120del), c.269T>C (p.Leu90Pro), c.235delC (p.Leu79Cysfs), c.229T>C (p.Trp77Arg), c.167delT (p.Leu56Argfs), c.109G>A (p.Val37Ile), c.71G>A (p.Trp24Ter), c.35delG (p.Gly12Valfs)

Cautions:

- The interpretation of this report is based upon the information provided by the ordering physician at the time of testing. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.
- This test is designed to detect the specific clinically validated variants listed in this report. It is possible that other types of disease-causing variants exist in this or other related genes that were not detected.
- Patients who have ever had an allogeneic blood or marrow transplant or who have received a heterologous blood transfusion within the last month, can have inaccurate genetic test results due to presence of donor DNA. Patients who have undergone liver transplantation may also have inaccurate genetic test results.
- Variant nomenclature is based on genomic build GRCh38. Variants are evaluated and classified according to ACMG recommendations (Richards, et al., 2015). Variant classification may change over time. If a variant that was part of Mayo Clinic GeneGuide™ is reclassified and you have questions, please contact us at 1-877-858-0398.

Cautions for Health Care Providers:

- This test is not intended to diagnose a disease, determine medical treatment, or tell the user anything about their current state of health.
- This test is intended to provide users with their genetic information to inform lifestyle decisions and conversations with their doctor or other healthcare professional.
- Any diagnostic or treatment decisions should be based on testing and/or other information that your healthcare provider determines to be appropriate for you.

Warnings:

- The test is intended only for autosomal recessive carrier screening in adults of reproductive age.
- The test does not detect all genetic variants related to the condition, and the absence of a variant tested does not rule out the presence of other genetic variants in this, or other genes, that may be disease related
- The results of this test are intended to be interpreted by a board-certified clinical molecular geneticist or equivalent and should be used in conjunction with other available laboratory and clinical information.
- This test is not intended for disease diagnosis, prenatal testing of fetuses, risk assessment, prognosis or pre-symptomatic testing, susceptibility testing, or newborn screening.
- This test is not intended to diagnose a disease, or tell you anything about your risk for developing a disease in the future. On its own, this test is also not intended to tell you anything about the health of your fetus, or your newborn child's risk of developing a particular disease later on in life.
- This test is not a substitute for visits to a healthcare provider. It is recommended that you consult with a healthcare provider if you have any questions or concerns about your results.
- The test does not diagnose any health conditions. Results should be used along with other clinical information for any medical purposes
- Your ethnicity may affect how your genetic health results are interpreted

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

MCAD Deficiency Results

Condition	Gene	# Variants	Variant(s) Identified	Variant(s) Classification	Interpretation
MCAD Deficiency	ACADM	0			No variants identified

MCAD Deficiency interpretation

We ruled out the most common genetic variants in the ACADM gene. Even though this test didn't identify any variants, there is a small chance that you still could be a carrier. The risk level depends in part on your ethnic background, as noted below in the residual risk information. For most people, there's nothing more you need to do based on this result. If you have a family history of MCAD deficiency, you may want to consider more comprehensive testing. Talk with your health care provider for information about comprehensive testing for MCAD deficiency.

Your body needs energy to function properly. Medium-chain acyl-CoA dehydrogenase deficiency (MCAD deficiency) is a genetic disease that affects your body's ability to break down certain fats and convert them into energy. When this process doesn't work properly, the level of sugar in your blood can become dangerously low. MCAD deficiency is an inherited genetic disease caused by variants in the ACADM gene located on chromosome 1. Most people with MCAD deficiency have two variants in the ACADM gene.

This test is not intended to diagnose a disease or tell you anything about your risk for developing a disease in the future. A genetic consultation may be of benefit. Please contact the independent genetic counseling provider who reviewed your test order and results, PWNHealth toll free at 1-888-494-7333 or by email at gc@pwnhealth.com, if you have questions regarding this report.

Residual Risk Information:

Chance of having a genetic variant based on this test:
- Non-Hispanic White (European): Less than 1 in 300 (0.33 percent)
- Hispanic White: Data are not available on how much risk is reduced
- African-American: Data are not available on how much risk is reduced
- Japanese/Asian: Data are not available on how much risk is reduced
- Native American: Data are not available on how much risk is reduced
- Other/Mixed race: Data are not available on how much risk is reduced

Method: The variants interpreted by the Mayo Clinic GeneGuide™ system are detected by our partner, Helix. The Helix Exome+ assay uses next generation sequencing (NGS) of DNA to analyze the entire human exome plus several hundred thousand additional informative regions outside of the exome, including the complete mitochondrial genome. The Helix Exome+ assay was performed on genomic DNA extracted from your submitted saliva sample. DNA from your sample was captured and enriched using a custom set of reagents for target selection. Next Generation Sequencing was performed on an Illumina DNA sequencing instrument using HapMap samples NA12878 and NA12877 as internal controls. Alignment to a modified version of GRCh38 and variant calling were completed using a customized version of Sentieon's DNAseq software. The Helix bioinformatics pipeline applied several quality control filters to produce the final passing variants list. Using a semi-automated platform for laboratory review and reporting, The Mayo Clinic GeneGuide™ interpretive system then pulled a predetermined set of clinically validated variants from the Helix Secure Database, and matched the variants to the appropriate result interpretation.

Variants Tested: The variant nomenclature that is used to describe the MCAD Deficiency variants included in Mayo Clinic GeneGuide™ is based on:

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

NM_000016.5(ACADM)

c.362C>T (p.Thr121Ile), c.449_452delCTGA (p.Thr150Argfs), c.799G>A (p.Gly267Arg), c.985A>G (p.Lys329Glu), c.1102_1105delTTAG (p.Ala369Leufs)

Cautions:

- The interpretation of this report is based upon the information provided by the ordering physician at the time of testing. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.
- This test is designed to detect the specific clinically validated variants listed in this report. It is possible that other types of disease-causing variants exist in this or other related genes that were not detected.
- Patients who have ever had an allogeneic blood or marrow transplant or who have received a heterologous blood transfusion within the last month, can have inaccurate genetic test results due to presence of donor DNA. Patients who have undergone liver transplantation may also have inaccurate genetic test results.
- Variant nomenclature is based on genomic build GRCh38. Variants are evaluated and classified according to ACMG recommendations (Richards, et al., 2015). Variant classification may change over time. If a variant that was part of Mayo Clinic GeneGuide™ is reclassified and you have questions, please contact us at 1-877-858-0398.

Cautions for Health Care Providers:

- This test is not intended to diagnose a disease, determine medical treatment, or tell the user anything about their current state of health.
- This test is intended to provide users with their genetic information to inform lifestyle decisions and conversations with their doctor or other healthcare professional.
- Any diagnostic or treatment decisions should be based on testing and/or other information that your healthcare provider determines to be appropriate for you.

Warnings:

- The test is intended only for autosomal recessive carrier screening in adults of reproductive age.
- The test does not detect all genetic variants related to the condition, and the absence of a variant tested does not rule out the presence of other genetic variants in this, or other genes, that may be disease related
- The results of this test are intended to be interpreted by a board-certified clinical molecular geneticist or equivalent and should be used in conjunction with other available laboratory and clinical information.
- This test is not intended for disease diagnosis, prenatal testing of fetuses, risk assessment, prognosis or pre-symptomatic testing, susceptibility testing, or newborn screening.
- This test is not intended to diagnose a disease, or tell you anything about your risk for developing a disease in the future. On its own, this test is also not intended to tell you anything about the health of your fetus, or your newborn child's risk of developing a particular disease later on in life.
- This test is not a substitute for visits to a healthcare provider. It is recommended that you consult with a healthcare provider if you have any questions or concerns about your results.
- The test does not diagnose any health conditions. Results should be used along with other clinical information for any medical purposes
- Your ethnicity may affect how your genetic health results are interpreted

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Sickle Cell Disease Results

Condition	Gene	# Variants	Variant(s) Identified	Variant(s) Classification	Interpretation
Sickle Cell Disease	HBB	0			No variants identified

Sickle Cell Disease interpretation

We did not detect either of the 2 variants (Hb S, Hb C) in the HBB gene associated with sickle cell disease. Even though this test didn't identify the sickle cell variants in the HBB gene, there is a chance that you could be a carrier of another HBB variant. The risk level depends in part on your ethnic background, as noted below in the residual risk information. For most people, there's nothing more you need to do based on this result.

Sickle cell disease is a condition in which normal red blood cells, which are flexible and round, become stiff and sickle-shaped. This happens because the hemoglobin molecules that deliver oxygen to the body are abnormal. Sickle cells break apart easily and do not live as long as normal red blood cells. This leads to a shortage of red blood cells, known as anemia. Sickle cell disease is an inherited genetic condition caused by variants in the HBB gene on chromosome 11. Most people with sickle cell disease have the Hb S variant on both of their HBB gene copies. However, there are other variants in the HBB gene that, when combined with Hb S, also cause the symptoms of sickle cell disease. A rare variant called Hb C is similar to Hb S but has a milder effect. People with one Hb S and one Hb C variant usually have a milder form of sickle cell disease. People with two Hb C variants have Hemoglobin C disease. Unlike sickle cell disease, most people with hemoglobin C disease do not have symptoms or medical problems. Some people with hemoglobin C disease have mild anemia and/or an enlarged spleen. Gallstones are more common. This test is not intended to diagnose a disease or tell you anything about your risk for developing a disease in the future.

A genetic consultation may be of benefit. Please contact the independent genetic counseling provider who reviewed your test order and results, PWNHealth toll free at 1-888-494-7333 or by email at gc@pwnhealth.com, if you have questions regarding this report.

Residual Risk Information:
- African American: 1 in 43 (2.3%)
- Middle Eastern: 1 in 84 (1.2%)
- Non-Hispanic White: 1 in 1200 (0.08%)
- Ashkenazi Jewish: 1 in 930 (0.1%)
- Other/Mixed Race: No specific data is available.

Method: The variants interpreted by the Mayo Clinic GeneGuide™ system are detected by our partner, Helix. The Helix Exome+ assay uses next generation sequencing (NGS) of DNA to analyze the entire human exome plus several hundred thousand additional informative regions outside of the exome, including the complete mitochondrial genome. The Helix Exome+ assay was performed on genomic DNA extracted from your submitted saliva sample. DNA from your sample was captured and enriched using a custom set of reagents for target selection. Next Generation Sequencing was performed on an Illumina DNA sequencing instrument using HapMap samples NA12878 and NA12877 as internal controls. Alignment to a modified version of GRCh38 and variant calling were completed using a customized version of Sentieon's DNAseq software. The Helix bioinformatics pipeline applied several quality control filters to produce the final passing variants list. Using a semi-automated platform for laboratory review and reporting, The Mayo Clinic GeneGuide™ interpretive system then pulled a predetermined set of clinically validated variants from the Helix Secure Database, and matched the variants to the appropriate result interpretation.

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Variants Tested: The variant nomenclature that is used to describe the Sickle Cell Disease variants included in Mayo Clinic GeneGuide™ is based on:

NM_000518.4(HBB)

c.20A>T (p.Glu7Val), c.19G>A (p.Glu7Lys)

Cautions:

- The interpretation of this report is based upon the information provided by the ordering physician at the time of testing. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.
- This test is designed to detect the specific clinically validated variants listed in this report. It is possible that other types of disease-causing variants exist in this or other related genes that were not detected.
- Patients who have ever had an allogeneic blood or marrow transplant or who have received a heterologous blood transfusion within the last month, can have inaccurate genetic test results due to presence of donor DNA. Patients who have undergone liver transplantation may also have inaccurate genetic test results.
- Variant nomenclature is based on genomic build GRCh38. Variants are evaluated and classified according to ACMG recommendations (Richards, et al., 2015). Variant classification may change over time. If a variant that was part of Mayo Clinic GeneGuide™ is reclassified and you have questions, please contact us at 1-877-858-0398.

Cautions for Health Care Providers:

- This test is not intended to diagnose a disease, determine medical treatment, or tell the user anything about their current state of health.
- This test is intended to provide users with their genetic information to inform lifestyle decisions and conversations with their doctor or other healthcare professional.
- Any diagnostic or treatment decisions should be based on testing and/or other information that your healthcare provider determines to be appropriate for you.

Warnings:

- The test is intended only for autosomal recessive carrier screening in adults of reproductive age.
- The test does not detect all genetic variants related to the condition, and the absence of a variant tested does not rule out the presence of other genetic variants in this, or other genes, that may be disease related
- The results of this test are intended to be interpreted by a board-certified clinical molecular geneticist or equivalent and should be used in conjunction with other available laboratory and clinical information.
- This test is not intended for disease diagnosis, prenatal testing of fetuses, risk assessment, prognosis or pre-symptomatic testing, susceptibility testing, or newborn screening.
- This test is not intended to diagnose a disease, or tell you anything about your risk for developing a disease in the future. On its own, this test is also not intended to tell you anything about the health of your fetus, or your newborn child's risk of developing a particular disease later on in life.
- This test is not a substitute for visits to a healthcare provider. It is recommended that you consult with a healthcare provider if you have any questions or concerns about your results.
- The test does not diagnose any health conditions. Results should be used along with other clinical information for any medical purposes
- Your ethnicity may affect how your genetic health results are interpreted

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Introduction to Medication Response

Some genes control the way your body processes medications and anesthesia, in the same way other genes determine things like your hair color and height. Like the genes that determine your physical traits, the genes that influence your response to medications/anesthesia vary from person to person. As a result, medications/anesthesia drugs don't affect everyone the same way. While most people respond to common drugs as expected, the same drugs may have little effect on a small subset of those who try them. Conversely, another subset may experience too much effect from particular medications, developing potentially serious side effects. In both cases, genetic variants may be the cause. These variants are likely to be in genes that code for enzymes needed to break medications down to a form that can reach and act on targeted organs or systems in the body.
If you have concerns about your response to particular medications, discuss them with your health care provider. Pharmacogenomic testing is one tool that can help your health care provider determine the best medication for you. Your health care provider also considers other factors such as your age, lifestyle, other medications you are taking and your overall health when choosing the right treatment for you.

Pharmacogenomic tests are only available for certain medications. If you are taking a medication for which pharmacogenomic testing is not available, you will not be able to determine how your genetic makeup will influence your response to that medication.

Remember, never change your medications without first discussing it with your health care provider.

Medication Response Result Summary

Condition	Gene	# Variants	Variant(s) Identified	Variant(s) Classification	Interpretation
Ibuprofen Metabolism	CYP2C9	1	c.1075A>C (p.Ile359Leu)	*1/*3	Intermediate metabolizer
Malignant Hyperthermia Susceptibility	CACNA1S	0			No variants identified
	RYR1	0			
Omeprazole Metabolism	CYP2C19	0			Normal metabolizer
Pseudocholinesterase Deficiency	BCHE	0			No variants identified

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Ibuprofen Metabolism Results

Condition	Gene	# Variants	Variant(s) Identified	Variant(s) Classification	Interpretation
Ibuprofen Metabolism	CYP2C9	1	c.1075A>C (p.Ile359Leu)	*1/*3	Intermediate metabolizer

Ibuprofen Metabolism interpretation

Based on your CYP2C9 result listed in the table above, you are likely an intermediate metabolizer. This means that you likely break down (metabolize) ibuprofen somewhat slowly and you may be at increased risk of side effects from ibuprofen.

Ibuprofen is a commonly used over-the-counter medication for treating mild to moderate pain and inflammation. It is available under a variety of brand names, including Advil, Motrin IB and others. The CYP2C9 gene provides instructions for cells to create an enzyme that breaks down (metabolizes) ibuprofen. Some variants in the CYP2C9 gene decrease ibuprofen metabolism causing the medication to build up in the body over time, which may increase the risk of side effects. People with these types of variants are identified as poor or intermediate metabolizers. This test is not intended to diagnose a disease or tell you anything about your risk for developing a disease in the future.

The CYP2C9 gene affects some prescription medications. Talk with your health care provider to learn if any of your prescription medications are affected. Before you make any changes to your medications, talk with your health care provider.A genetic consultation may be of benefit. Please contact the independent genetic counseling provider who reviewed your test order and results, PWNHealth toll free at 1-888-494-7333 or by email at gc@pwnhealth.com, if you have questions regarding this report.

Method: The variants interpreted by the Mayo Clinic GeneGuide™ system are detected by our partner, Helix. The Helix Exome+ assay uses next generation sequencing (NGS) of DNA to analyze the entire human exome plus several hundred thousand additional informative regions outside of the exome, including the complete mitochondrial genome. The Helix Exome+ assay was performed on genomic DNA extracted from your submitted saliva sample. DNA from your sample was captured and enriched using a custom set of reagents for target selection. Next Generation Sequencing was performed on an Illumina DNA sequencing instrument using HapMap samples NA12878 and NA12877 as internal controls. Alignment to a modified version of GRCh38 and variant calling were completed using a customized version of Sentieon's DNAseq software. The Helix bioinformatics pipeline applied several quality control filters to produce the final passing variants list. Using a semi-automated platform for laboratory review and reporting, The Mayo Clinic GeneGuide™ interpretive system then pulled a predetermined set of clinically validated variants from the Helix Secure Database, and matched the variants to the appropriate result interpretation.

Variants Tested: The variant nomenclature that is used to describe the Ibuprofen Metabolism variants included in Mayo Clinic GeneGuide™ is based on:

NM_000771.3(CYP2C9)

c.430C>T (p.Arg144Cys), c.1075A>C (p.Ile359Leu)

Cautions:

- The interpretation of this report is based upon the information provided by the ordering physician at the time of testing. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.
- This test is designed to detect the specific clinically validated variants listed in this report. It is possible that other types of disease-causing variants exist in this or other related genes that were not detected.
- Patients who have ever had an allogeneic blood or marrow transplant or who have received a heterologous blood transfusion within the last month, can have inaccurate genetic test results due to presence of donor DNA. Patients who have undergone liver

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

transplantation may also have inaccurate genetic test results.
- Variant nomenclature is based on genomic build GRCh38. Variants are evaluated and classified according to ACMG recommendations (Richards, et al., 2015). Variant classification may change over time. If a variant that was part of Mayo Clinic GeneGuide™ is reclassified and you have questions, please contact us at 1-877-858-0398.

Cautions for Health Care Providers:

- This test is not intended to diagnose a disease, determine medical treatment, or tell the user anything about their current state of health.
- This test is intended to provide users with their genetic information to inform lifestyle decisions and conversations with their doctor or other healthcare professional.
- Any diagnostic or treatment decisions should be based on testing and/or other information that your healthcare provider determines to be appropriate for you.

Warnings:

- This test is not intended to diagnose a disease, tell you anything about your current state of health, or be used to make medical decisions, including whether or not you should take a medication or how much of a medication you should take.
- Do NOT adjust dosage of ANY medication (including ibuprofen or omeprazole) without consulting a health care provider.
- This test reports information about CYP2C9 and CYP2C19 variants as they relate to ibuprofen and omeprazole metabolism ONLY. This test is not designed to provide information about metabolism of other drugs that might be associated with CYP2C9 and CYP2C19.
- This test provides genetic risk information based on assessment of specific genetic variants but does not report on a user's entire genetic profile.
- This test does not detect all genetic variants related to a given disease, and the absence of a variant tested does not rule out the presence of other genetic variants that may be related to the disease.
- Other companies offering a genetic risk test may be detecting different genetic variants for the same disease, so the user may get different results using a test from a different company.
- Factors such as environmental and lifestyle risk factors may affect the risk of developing a given disease.
- Some people may feel anxious about getting genetic test health results. This is normal. If the potential user feels very anxious, such user should speak to his or her doctor or other healthcare professional prior to collection of a sample for testing. This test is not a substitute for visits to a doctor or other healthcare professional. Users should consult with their doctor or other healthcare professional if they have any questions or concerns about the results of their test or their current state of health.

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Malignant Hyperthermia Susceptibility Results

Condition	Gene	# Variants	Variant(s) Identified	Variant(s) Classification	Interpretation
Malignant Hyperthermia Susceptibility	CACNA1S	0			No variants identified
	RYR1	0			

Malignant Hyperthermia Susceptibility interpretation

This test did not identify variants in the RYR1 or CACNA1S genes. You are not likely to have complications from anesthesia drugs associated with malignant hyperthermia. Even though this test didn't identify any variants, there is a chance that you still could have a different variant. If someone in your family has malignant hyperthermia susceptibility or has serious side effects with anesthesia, make sure to tell your healthcare provider. If you have a personal history OR family history of malignant hyperthermia, your risk may still be increased. Talk with your health care provider about your risk.

Malignant hyperthermia susceptibility is a rare condition that increases the risk of having a severe reaction to certain drugs used as part of anesthesia for surgery. The reaction, called malignant hyperthermia, includes a high body temperature, fever, breakdown of muscle fibers, increased acid in the blood, fast breathing and fast heartbeat. Without proper treatment, malignant hyperthermia can be fatal. Most of the time, people with malignant hyperthermia susceptibility will only have symptoms when they are exposed to anesthesia medications that trigger the abnormal calcium release in muscle cells. Rarely, extreme heat or exercise can trigger the symptoms as well. Malignant hyperthermia susceptibility is passed down in families in an autosomal dominant pattern, so people with one variant are considered affected. Variations in two different genes that affect calcium levels (RYR1 and CACNA1S) increase risk for malignant hyperthermia susceptibility. The RYR1 gene is located on chromosome 19, and the CACNA1S gene is located on chromosome 1.

This test is not intended to diagnose a disease or tell you anything about your risk for developing a disease in the future. This test result does not mean you'll have a normal response to all medications. Talk with your health care provider if you have concerns about your response to other medications. Before you make any changes to your medications, talk with your health care provider.

A genetic consultation may be of benefit. Please contact the independent genetic counseling provider who reviewed your test order and results, PWNHealth toll free at 1-888-494-7333 or by email at gc@pwnhealth.com, if you have questions regarding this report.

Method: The variants interpreted by the Mayo Clinic GeneGuide™ system are detected by our partner, Helix. The Helix Exome+ assay uses next generation sequencing (NGS) of DNA to analyze the entire human exome plus several hundred thousand additional informative regions outside of the exome, including the complete mitochondrial genome. The Helix Exome+ assay was performed on genomic DNA extracted from your submitted saliva sample. DNA from your sample was captured and enriched using a custom set of reagents for target selection. Next Generation Sequencing was performed on an Illumina DNA sequencing instrument using HapMap samples NA12878 and NA12877 as internal controls. Alignment to a modified version of GRCh38 and variant calling were completed using a customized version of Sentieon's DNAseq software. The Helix bioinformatics pipeline applied several quality control filters to produce the final passing variants list. Using a semi-automated platform for laboratory review and reporting, The Mayo Clinic GeneGuide™ interpretive system then pulled a predetermined set of clinically validated variants from the Helix Secure Database, and matched the variants to the appropriate result interpretation.

Variants Tested: The variant nomenclature that is used to describe the Malignant Hyperthermia Susceptibility variants included in Mayo Clinic GeneGuide™ is based on:

NM_000069.2(CACNA1S)

c.3257G>A (p.Arg1086His), c.520C>T (Arg174Trp)

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

NM_000540.2(RYR1)

c.103T>C (p.Cys35Arg), c.487C>T (p.Arg163Cys), c.488G>T (p.Arg163Leu), c.742G>A (p.Gly248Arg), c.742G>C (p.Gly248Arg), c.1021G>A (p.Gly341Arg), c.1021G>C (p.Gly341Arg), c.1201C>T (p.Arg401Cys), c.1209C>G (p.Ile403Met), c.1565A>C (p.Tyr522Ser), c.1589G>A (p.Arg530His), c.1654C>T (p.Arg552Trp), c.1840C>T (p.Arg614Cys), c.1841G>T (p.Arg614Leu), c.6487C>T (p.Arg2163Cys), c.6488G>A (p.Arg2163His), c.6502G>A (p.Val2168Met), c.6617C>G (p.Thr2206Arg), c.7007G>A (p.Arg2336His), c.7048G>A (p.Ala2350Thr), c.7124G>C (p.Gly2375Ala), c.7282G>A (p.Ala2428Thr), c.7300G>A (p.Gly2434Arg), c.7304G>A (p.Arg2435His), c.7360C>T (p.Arg2454Cys), c.7361G>A (p.Arg2454His), c.7372C>T (p.Arg2458Cys), c.7373G>A (p.Arg2458His), c.7522C>T (p.Arg2508Cys), c.7523G>A (p.Arg2508His), c.14387A>G (p.Tyr4796Cys), c.14477C>T (p.Thr4826Ile), c.14497C>T (p.His4833Tyr), c.14512C>G (p.Leu4838Val), c.14545G>A (p.Val4849Ile), c.14582G>A (p.Arg4861His), c.14693T>C (p.Ile4898Thr)

Cautions:

- The interpretation of this report is based upon the information provided by the ordering physician at the time of testing. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.
- This test is designed to detect the specific clinically validated variants listed in this report. It is possible that other types of disease-causing variants exist in this or other related genes that were not detected.
- Patients who have ever had an allogeneic blood or marrow transplant or who have received a heterologous blood transfusion within the last month, can have inaccurate genetic test results due to presence of donor DNA. Patients who have undergone liver transplantation may also have inaccurate genetic test results.
- Variant nomenclature is based on genomic build GRCh38. Variants are evaluated and classified according to ACMG recommendations (Richards, et al., 2015). Variant classification may change over time. If a variant that was part of Mayo Clinic GeneGuide™ is reclassified and you have questions, please contact us at 1-877-858-0398.

Cautions for Health Care Providers:

- This test is not intended to diagnose a disease, determine medical treatment, or tell the user anything about their current state of health.
- This test is intended to provide users with their genetic information to inform lifestyle decisions and conversations with their doctor or other healthcare professional.
- Any diagnostic or treatment decisions should be based on testing and/or other information that your healthcare provider determines to be appropriate for you.

Warnings:

- This test is not intended to diagnose a disease, tell you anything about your current state of health, or be used to make medical decisions, including whether or not you should take a medication or how much of a medication you should take.
- Do NOT adjust dosage of ANY medication (including ibuprofen or omeprazole) without consulting a health care provider.
- This test reports information about CYP2C9 and CYP2C19 variants as they relate to ibuprofen and omeprazole metabolism ONLY. This test is not designed to provide information about metabolism of other drugs that might be associated with CYP2C9 and CYP2C19.
- This test provides genetic risk information based on assessment of specific genetic variants but does not report on a user's entire genetic profile.
- This test does not detect all genetic variants related to a given disease, and the absence of a variant tested does not rule out the presence of other genetic variants that may be related to the disease.
- Other companies offering a genetic risk test may be detecting different genetic variants for the same disease, so the user may get different results using a test from a different company.
- Factors such as environmental and lifestyle risk factors may affect the risk of developing a given disease.
- Some people may feel anxious about getting genetic test health results. This is normal. If the potential user feels very anxious, such user should speak to his or her doctor or other healthcare professional prior to collection of a sample for testing. This test is not a substitute for visits to a doctor or other healthcare professional. Users should consult with their doctor or other healthcare

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

professional if they have any questions or concerns about the results of their test or their current state of health.

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Omeprazole Metabolism Results

Condition	Gene	# Variants	Variant(s) Identified	Variant(s) Classification	Interpretation
Omeprazole Metabolism	CYP2C19	0			Normal metabolizer

Omeprazole Metabolism interpretation

This test did not identify any variants in the CYP2C19 gene. You likely break down (metabolize) omeprazole normally and are likely not at increased risk for side effects from omeprazole. There is a small chance that you carry an uncommon genetic change that this test is not designed to detect.

Omeprazole is a medication used to relieve abdominal pain such as heartburn by reducing stomach acids. It's available by prescription or over-the-counter (OTC) under a variety of brand names, including Prilosec OTC, Zegerid OTC and others. The CYP2C19 gene is associated with omeprazole metabolism and is located on chromosome 10. The CYP2C19 gene provides instructions for cells to create an enzyme which breaks down (metabolizes) omeprazole. Some variants in the CYP2C19 gene decrease omeprazole metabolism causing the medication to build up in the body over time which may increase the risk for side effects. People with these types of variants are identified as poor or intermediate metabolizers. Other variants break down (metabolize) omeprazole quickly. People with these types of variants are known as ultrarapid metabolizers. Some variants in the CYP2C19 gene increase omeprazole metabolism causing rapid removal of the medication. This may decrease the effectiveness of omeprazole. People with these types of variants are identified as ultrarapid metabolizers. This test is not intended to diagnose a disease or tell you anything about your risk for developing a disease in the future. This test result does not mean you'll have a normal response to all medications. Talk with your health care provider if you have concerns about your response to medications.

Before you make any changes to your medications, talk with your health care provider.

A genetic consultation may be of benefit. Please contact the independent genetic counseling provider who reviewed your test order and results, PWNHealth toll free at 1-888-494-7333 or by email at gc@pwnhealth.com, if you have questions regarding this report.

Method: The variants interpreted by the Mayo Clinic GeneGuide™ system are detected by our partner, Helix. The Helix Exome+ assay uses next generation sequencing (NGS) of DNA to analyze the entire human exome plus several hundred thousand additional informative regions outside of the exome, including the complete mitochondrial genome. The Helix Exome+ assay was performed on genomic DNA extracted from your submitted saliva sample. DNA from your sample was captured and enriched using a custom set of reagents for target selection. Next Generation Sequencing was performed on an Illumina DNA sequencing instrument using HapMap samples NA12878 and NA12877 as internal controls. Alignment to a modified version of GRCh38 and variant calling were completed using a customized version of Sentieon's DNAseq software. The Helix bioinformatics pipeline applied several quality control filters to produce the final passing variants list. Using a semi-automated platform for laboratory review and reporting, The Mayo Clinic GeneGuide™ interpretive system then pulled a predetermined set of clinically validated variants from the Helix Secure Database, and matched the variants to the appropriate result interpretation.

Variants Tested: The variant nomenclature that is used to describe the Omeprazole Metabolism variants included in Mayo Clinic GeneGuide™ is based on:

NM_000769.2(CYP2C19)

c.-806C>T, c.636G>A (p.Trp212Ter), c.681G>A (p.Pro227=)

Cautions:

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

- The interpretation of this report is based upon the information provided by the ordering physician at the time of testing. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.
- This test is designed to detect the specific clinically validated variants listed in this report. It is possible that other types of disease-causing variants exist in this or other related genes that were not detected.
- Patients who have ever had an allogeneic blood or marrow transplant or who have received a heterologous blood transfusion within the last month, can have inaccurate genetic test results due to presence of donor DNA. Patients who have undergone liver transplantation may also have inaccurate genetic test results.
- Variant nomenclature is based on genomic build GRCh38. Variants are evaluated and classified according to ACMG recommendations (Richards, et al., 2015). Variant classification may change over time. If a variant that was part of Mayo Clinic GeneGuide™ is reclassified and you have questions, please contact us at 1-877-858-0398.

Cautions for Health Care Providers:

- This test is not intended to diagnose a disease, determine medical treatment, or tell the user anything about their current state of health.
- This test is intended to provide users with their genetic information to inform lifestyle decisions and conversations with their doctor or other healthcare professional.
- Any diagnostic or treatment decisions should be based on testing and/or other information that your healthcare provider determines to be appropriate for you.

Warnings:

- This test is not intended to diagnose a disease, tell you anything about your current state of health, or be used to make medical decisions, including whether or not you should take a medication or how much of a medication you should take.
- Do NOT adjust dosage of ANY medication (including ibuprofen or omeprazole) without consulting a health care provider.
- This test reports information about CYP2C9 and CYP2C19 variants as they relate to ibuprofen and omeprazole metabolism ONLY. This test is not designed to provide information about metabolism of other drugs that might be associated with CYP2C9 and CYP2C19.
- This test provides genetic risk information based on assessment of specific genetic variants but does not report on a user's entire genetic profile.
- This test does not detect all genetic variants related to a given disease, and the absence of a variant tested does not rule out the presence of other genetic variants that may be related to the disease.
- Other companies offering a genetic risk test may be detecting different genetic variants for the same disease, so the user may get different results using a test from a different company.
- Factors such as environmental and lifestyle risk factors may affect the risk of developing a given disease.
- Some people may feel anxious about getting genetic test health results. This is normal. If the potential user feels very anxious, such user should speak to his or her doctor or other healthcare professional prior to collection of a sample for testing. This test is not a substitute for visits to a doctor or other healthcare professional. Users should consult with their doctor or other healthcare professional if they have any questions or concerns about the results of their test or their current state of health.

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Pseudocholinesterase Deficiency Results

Condition	Gene	# Variants	Variant(s) Identified	Variant(s) Classification	Interpretation
Pseudocholinesterase Deficiency	BCHE	0			No variants identified

Pseudocholinesterase Deficiency interpretation

No variants in the BCHE gene were detected. In absence of nongenetic factors, you are not likely to be at increased risk of side effects from medications broken down by pseudocholinesterase. However, there is a chance that you carry an uncommon genetic variant in BCHE that this test isn't designed to detect. If you have a personal or family history of pseudocholinesterase deficiency, talk to your health care provider.

Pseudocholinesterase deficiency is a condition that makes a person sensitive to certain drugs used during anesthesia. Pseudocholinesterase (pronounced "soo-doe-coe-lin-es-ter-ase") is an enzyme found in the blood that is produced in the liver. It breaks down chemicals known as choline esters. A common drug made from choline esters is succinylcholine, which is used to relax muscles during some medical and surgical procedures. A person with pseudocholinesterase deficiency has less effective enzyme for processing succinylcholine. In this case, muscles stay relaxed for longer than expected. This can prevent the person from moving or breathing on his or her own, requiring breathing assistance for longer than expected. Pseudocholinesterase deficiency can be due to inheritance of variants in the BCHE gene on chromosome 3. Pseudocholinesterase deficiency can also be caused by other medical situations, such as pregnancy, liver disease or kidney disease. Inherited pseudocholinesterase deficiency is an autosomal recessive condition, so most affected people have two variants in the BCHE gene. However, to be cautious, health care providers prescribing anesthesia will likely opt for a medication not broken down by pseudocholinesterase in someone in which just one variant in BCHE is found.

This test is not intended to diagnose a disease or tell you anything about your risk for developing a disease in the future. This test result does not mean you'll have a normal response to all medications. Talk with your health care provider if you have concerns about your response to other medications.

Before you make any changes to your medications, talk with your health care provider.

A genetic consultation may be of benefit. Please contact the independent genetic counseling provider who reviewed your test order and results, PWNHealth toll free at 1-888-494-7333 or by email at gc@pwnhealth.com, if you have questions regarding this report.

Method: The variants interpreted by the Mayo Clinic GeneGuide™ system are detected by our partner, Helix. The Helix Exome+ assay uses next generation sequencing (NGS) of DNA to analyze the entire human exome plus several hundred thousand additional informative regions outside of the exome, including the complete mitochondrial genome. The Helix Exome+ assay was performed on genomic DNA extracted from your submitted saliva sample. DNA from your sample was captured and enriched using a custom set of reagents for target selection. Next Generation Sequencing was performed on an Illumina DNA sequencing instrument using HapMap samples NA12878 and NA12877 as internal controls. Alignment to a modified version of GRCh38 and variant calling were completed using a customized version of Sentieon's DNAseq software. The Helix bioinformatics pipeline applied several quality control filters to produce the final passing variants list. Using a semi-automated platform for laboratory review and reporting, The Mayo Clinic GeneGuide™ interpretive system then pulled a predetermined set of clinically validated variants from the Helix Secure Database, and matched the variants to the appropriate result interpretation.

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Variants Tested: The variant nomenclature that is used to describe the Pseudocholinesterase Deficiency variants included in Mayo Clinic GeneGuide™ is based on:

NM_000055.3(BCHE)

c.1574A>T (p.Glu525Val), c.1072T>A (p.Leu358Ile), c.1004T>C (p.Leu335Pro), c.812C>T (p.Thr271Met), c.467A>G (p.Tyr156Cys), c.293A>G (p.Asp98Gly)

NM_000055.2(BCHE)

c.435delTinsAG (p.Phe146Valfs)

Cautions:

- The interpretation of this report is based upon the information provided by the ordering physician at the time of testing. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.
- This test is designed to detect the specific clinically validated variants listed in this report. It is possible that other types of disease-causing variants exist in this or other related genes that were not detected.
- Patients who have ever had an allogeneic blood or marrow transplant or who have received a heterologous blood transfusion within the last month, can have inaccurate genetic test results due to presence of donor DNA. Patients who have undergone liver transplantation may also have inaccurate genetic test results.
- Variant nomenclature is based on genomic build GRCh38. Variants are evaluated and classified according to ACMG recommendations (Richards, et al., 2015). Variant classification may change over time. If a variant that was part of Mayo Clinic GeneGuide™ is reclassified and you have questions, please contact us at 1-877-858-0398.

Cautions for Health Care Providers:

- This test is not intended to diagnose a disease, determine medical treatment, or tell the user anything about their current state of health.
- This test is intended to provide users with their genetic information to inform lifestyle decisions and conversations with their doctor or other healthcare professional.
- Any diagnostic or treatment decisions should be based on testing and/or other information that your healthcare provider determines to be appropriate for you.

Warnings:

- This test is not intended to diagnose a disease, tell you anything about your current state of health, or be used to make medical decisions, including whether or not you should take a medication or how much of a medication you should take.
- Do NOT adjust dosage of ANY medication (including ibuprofen or omeprazole) without consulting a health care provider.
- This test reports information about CYP2C9 and CYP2C19 variants as they relate to ibuprofen and omeprazole metabolism ONLY. This test is not designed to provide information about metabolism of other drugs that might be associated with CYP2C9 and CYP2C19.
- This test provides genetic risk information based on assessment of specific genetic variants but does not report on a user's entire genetic profile.
- This test does not detect all genetic variants related to a given disease, and the absence of a variant tested does not rule out the presence of other genetic variants that may be related to the disease.
- Other companies offering a genetic risk test may be detecting different genetic variants for the same disease, so the user may get different results using a test from a different company.
- Factors such as environmental and lifestyle risk factors may affect the risk of developing a given disease.
- Some people may feel anxious about getting genetic test health results. This is normal. If the potential user feels very anxious, such user should speak to his or her doctor or other healthcare professional prior to collection of a sample for testing. This test is not a substitute for visits to a doctor or other healthcare professional. Users should consult with their doctor or other healthcare professional if they have any questions or concerns about the results of their test or their current state of health.

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Introduction to Disease Risk

For many diseases, inheriting one or more gene variants accounts for only part of your risk; non-genetic factors like environment or lifestyle also contribute to your risk. These types of conditions or diseases are referred to as complex or multifactorial, because genetic and non-genetic factors interact to contribute to risk.

Many common conditions are multifactorial, like depression, diabetes, high blood pressure, obesity and others. According to researchers, almost every disease has some genetic component. In some cases, genes play a large role in determining your risk, but in other cases genes have relatively little influence on risk. In those cases, environment, aging, and lifestyle factors can also increase your risk for disease, sometimes to a larger extent than genetic factors. Remember that in most cases, genes are not destiny. You can often lower your risk by leading a healthy lifestyle.

Having a genetic predisposition to a particular condition means that based on your genetic makeup, you are at higher than average risk of developing the condition. It doesn't mean that you will definitely develop that condition; just that you are at increased risk based on your genes and perhaps also based on your environment and lifestyle.

Disease Risk Result Summary

Condition	Gene	# Variants	Variant(s) Identified	Variant(s) Classification	Interpretation
Atrial Fibrillation	no-gene	0			Slightly increased risk
	ZFHX3	1	c.-24+40809G>A	Risk allele	
Age Related Macular Degeneration	CFH	0			Increased risk
	ARMS2	1	c.205G>T (p.Ala69Ser)	Risk allele	
Coronary Artery Disease	SORT1	1	c.*1859C>T	Risk allele	Slightly increased risk
	LPA	0			
	no-gene	2	g.22098575A>G	Risk allele	
Venous Thromboembolism	F5	1	c.1601G>A (p.Arg534Gln)	Risk allele	Increased risk
	F2	0			

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Atrial Fibrillation Results

Condition	Gene	# Variants	Variant(s) Identified	Variant(s) Classification	Interpretation
Atrial Fibrillation	no-gene	0			Slightly increased risk
	ZFHX3	1	c.-24+40809G>A	Risk allele	

Atrial Fibrillation interpretation

This test found one or more variants associated with a slightly increased risk of atrial fibrillation disease. People with these atrial fibrillation variants have a slightly increased risk of atrial fibrillation disease. These variants play only a minor role in your risk of atrial fibrillation. Lifestyle factors and family history also play an important role. Additionally, the variants tested for in Mayo Clinic GeneGuide™ are very common in the population.

Atrial fibrillation is an irregular and often rapid heart rate. It occurs when the heart's upper two chambers (the atria) beat irregularly and out of coordination with the two lower chambers of the heart. Atrial fibrillation can increase your risk of stroke, heart failure and other heart-related complications. It can also lead to blood clots forming in the heart that may travel to other organs and block blood flow. A very small number of families have a rare single gene variant causing them to be at high risk of atrial fibrillation. However, in most people, the genetic risk associated with atrial fibrillation comes from having one or more of the many genetic variants that only increase risk slightly. Two variants associated with atrial fibrillation: rs2106261 associated with ZFHX3 on chromosome 16 and rs2200733, an intergenic (no-gene) variant on chromosome 4. Having a variant does not mean that you, or your children, will develop atrial fibrillation. As a multifactorial condition, lifestyle factors also contribute to your risk of developing atrial fibrillation.

This test is not intended to diagnose a disease or tell you anything about your risk for developing a disease in the future. Talk with your health care provider for more information if any of the nongenetic factors indicate you are at increased risk.

A genetic consultation may be of benefit. Please contact the independent genetic counseling provider who reviewed your test order and results, PWNHealth toll free at 1-888-494-7333 or by email at gc@pwnhealth.com, if you have questions regarding this report.

Method: The variants interpreted by the Mayo Clinic GeneGuide™ system are detected by our partner, Helix. The Helix Exome+ assay uses next generation sequencing (NGS) of DNA to analyze the entire human exome plus several hundred thousand additional informative regions outside of the exome, including the complete mitochondrial genome. The Helix Exome+ assay was performed on genomic DNA extracted from your submitted saliva sample. DNA from your sample was captured and enriched using a custom set of reagents for target selection. Next Generation Sequencing was performed on an Illumina DNA sequencing instrument using HapMap samples NA12878 and NA12877 as internal controls. Alignment to a modified version of GRCh38 and variant calling were completed using a customized version of Sentieon's DNAseq software. The Helix bioinformatics pipeline applied several quality control filters to produce the final passing variants list. Using a semi-automated platform for laboratory review and reporting, The Mayo Clinic GeneGuide™ interpretive system then pulled a predetermined set of clinically validated variants from the Helix Secure Database, and matched the variants to the appropriate result interpretation.

Variants Tested: The variant nomenclature that is used to describe the Atrial Fibrillation variants included in Mayo Clinic GeneGuide™ is based on:

NC_000004.12

g.110789013C>T

NM_001164766.1(ZFHX3)

c.-24+40809G>A

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Cautions:

- The interpretation of this report is based upon the information provided by the ordering physician at the time of testing. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.
- This test is designed to detect the specific clinically validated variants listed in this report. It is possible that other types of disease-causing variants exist in this or other related genes that were not detected.
- Patients who have ever had an allogeneic blood or marrow transplant or who have received a heterologous blood transfusion within the last month, can have inaccurate genetic test results due to presence of donor DNA. Patients who have undergone liver transplantation may also have inaccurate genetic test results.
- Variant nomenclature is based on genomic build GRCh38. Variants are evaluated and classified according to ACMG recommendations (Richards, et al., 2015). Variant classification may change over time. If a variant that was part of Mayo Clinic GeneGuide™ is reclassified and you have questions, please contact us at 1-877-858-0398.

Cautions for Health Care Providers:

- This test is not intended to diagnose a disease, determine medical treatment, or tell the user anything about their current state of health.
- This test is intended to provide users with their genetic information to inform lifestyle decisions and conversations with their doctor or other healthcare professional.
- Any diagnostic or treatment decisions should be based on testing and/or other information that your healthcare provider determines to be appropriate for you.

Warnings:

- This test provides genetic risk information based on assessment of specific genetic variants but does not report on a user's entire genetic profile.
- This test does not detect all genetic variants related to a given disease, and the absence of a variant tested does not rule out the presence of other genetic variants that may be related to the disease
- Other companies offering a genetic risk test may be detecting different genetic variants for the same disease, so the user may get different results using a test from a different company.
- Factors such as environmental and lifestyle risk factors may affect the risk of developing a given disease.
- Some people may feel anxious about getting genetic test health results. This is normal. If the potential user feels very anxious, such user should speak to his or her doctor or other healthcare professional prior to collection of a sample for testing. This test is not a substitute for visits to a doctor or other healthcare professional. Users should consult with their doctor or other healthcare professional if they have any questions or concerns about the results of their test or their current state of health.
- This test is not intended to diagnose a disease, tell you anything about your current state of health, or be used to make medical decisions, including whether or not you should take a medication or how much of a medication you should take.

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Age Related Macular Degeneration Results

Condition	Gene	# Variants	Variant(s) Identified	Variant(s) Classification	Interpretation
Age Related Macular Degeneration	CFH	0			Increased risk
	ARMS2	1	c.205G>T (p.Ala69Ser)	Risk allele	

Age Related Macular Degeneration interpretation

This test found one or more variants associated with an increased risk of age-related macular degeneration. People with these ARMD variants have an increased risk of age-related macular degeneration. Lifestyle factors and family history also play an important role in your risk of ARMD. Additionally, these variants are very common in the population.

Age-related macular degeneration (ARMD) is a common eye disorder caused by damage to the macula in people over the age of 50. ARMD does not lead to complete loss of vision (blindness), but the loss of central vision may inhibit everyday activities. In some people, loss of vision occurs rapidly, while in others ARMD progression is slow enough that vision loss does not occur for many years. ARMD is caused by a combination of genetic and non-genetic factors. Some, but not all, of these factors are known. Several genes have been found to play a role in keeping the macula of the eye healthy. Two of these genes are called CFH on chromosome 1 and ARMS2 on chromosome 10. Mayo Clinic GeneGuide™ tests for two common genetic variants, rs1061170 in CFH and rs10490924 in ARMS2.

Having a variant does not mean that you, or your children, will develop ARMD. As a multifactorial condition, lifestyle factors also contribute to your risk of developing ARMD. This test is not intended to diagnose a disease or tell you anything about your risk for developing a disease in the future. If you are concerned about your risk of ARMD, talk to your health care provider about modifiable risk factors.

A genetic consultation may be of benefit. Please contact the independent genetic counseling provider who reviewed your test order and results, PWNHealth toll free at 1-888-494-7333 or by email at gc@pwnhealth.com, if you have questions regarding this report.

Method: The variants interpreted by the Mayo Clinic GeneGuide™ system are detected by our partner, Helix. The Helix Exome+ assay uses next generation sequencing (NGS) of DNA to analyze the entire human exome plus several hundred thousand additional informative regions outside of the exome, including the complete mitochondrial genome. The Helix Exome+ assay was performed on genomic DNA extracted from your submitted saliva sample. DNA from your sample was captured and enriched using a custom set of reagents for target selection. Next Generation Sequencing was performed on an Illumina DNA sequencing instrument using HapMap samples NA12878 and NA12877 as internal controls. Alignment to a modified version of GRCh38 and variant calling were completed using a customized version of Sentieon's DNAseq software. The Helix bioinformatics pipeline applied several quality control filters to produce the final passing variants list. Using a semi-automated platform for laboratory review and reporting, The Mayo Clinic GeneGuide™ interpretive system then pulled a predetermined set of clinically validated variants from the Helix Secure Database, and matched the variants to the appropriate result interpretation.

Variants Tested: The variant nomenclature that is used to describe the Age Related Macular Degeneration variants included in Mayo Clinic GeneGuide™ is based on:

NM_000186.3(CFH)

c.1204C= (p.His402=)

NM_001099667.1(ARMS2)

c.205G>T (p.Ala69Ser)

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Cautions:

- The interpretation of this report is based upon the information provided by the ordering physician at the time of testing. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.
- This test is designed to detect the specific clinically validated variants listed in this report. It is possible that other types of disease-causing variants exist in this or other related genes that were not detected.
- Patients who have ever had an allogeneic blood or marrow transplant or who have received a heterologous blood transfusion within the last month, can have inaccurate genetic test results due to presence of donor DNA. Patients who have undergone liver transplantation may also have inaccurate genetic test results.
- Variant nomenclature is based on genomic build GRCh38. Variants are evaluated and classified according to ACMG recommendations (Richards, et al., 2015). Variant classification may change over time. If a variant that was part of Mayo Clinic GeneGuide™ is reclassified and you have questions, please contact us at 1-877-858-0398.

Cautions for Health Care Providers:

- This test is not intended to diagnose a disease, determine medical treatment, or tell the user anything about their current state of health.
- This test is intended to provide users with their genetic information to inform lifestyle decisions and conversations with their doctor or other healthcare professional.
- Any diagnostic or treatment decisions should be based on testing and/or other information that your healthcare provider determines to be appropriate for you.

Warnings:

- This test provides genetic risk information based on assessment of specific genetic variants but does not report on a user's entire genetic profile.
- This test does not detect all genetic variants related to a given disease, and the absence of a variant tested does not rule out the presence of other genetic variants that may be related to the disease
- Other companies offering a genetic risk test may be detecting different genetic variants for the same disease, so the user may get different results using a test from a different company.
- Factors such as environmental and lifestyle risk factors may affect the risk of developing a given disease.
- Some people may feel anxious about getting genetic test health results. This is normal. If the potential user feels very anxious, such user should speak to his or her doctor or other healthcare professional prior to collection of a sample for testing. This test is not a substitute for visits to a doctor or other healthcare professional. Users should consult with their doctor or other healthcare professional if they have any questions or concerns about the results of their test or their current state of health.
- This test is not intended to diagnose a disease, tell you anything about your current state of health, or be used to make medical decisions, including whether or not you should take a medication or how much of a medication you should take.

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Coronary Artery Disease Results

Condition	Gene	# Variants	Variant(s) Identified	Variant(s) Classification	Interpretation
Coronary Artery Disease	SORT1	1	c.*1859C>T	Risk allele	Slightly increased risk
	LPA	0			
	no-gene	2	g.22098575A>G	Risk allele	

Coronary Artery Disease interpretation

This test found one or more variants associated with a slightly increased risk of coronary artery disease. People with these CAD variants have a slightly increased risk of coronary artery disease. These variants play only a minor role in your risk of coronary artery disease. Lifestyle factors and family history also play an important role. Additionally, the variants we test for are very common in the population.

Coronary artery disease is the major cause of heart attacks. It affects the large blood vessels (coronary arteries) that supply the heart with oxygen-rich blood and nutrients. Coronary artery disease occurs when the coronary arteries become narrowed or blocked by a fatty cholesterol-containing substance called plaque. Over time the buildup of plaque causes the arteries to harden and narrow. This makes it difficult for blood to flow through the body. Signs and symptoms include shortness of breath, chest pain and heart attack. These may not occur until plaque has built up for many years. Research suggests that about 50 percent of coronary artery disease risk is due to genetic factors. Many genetic variants that increase risk have been identified. Each of these variants only explains a small part of the genetic risk of coronary artery disease. Mayo Clinic GeneGuide™ tests for three of these variants: rs4977574 an intergenic (no-gene) variant located on chromosome 9, rs3798220 associated with LPA on chromosome 6, and rs646776 associated with SORT1 on chromosome 1. Having a variant does not mean that you, or your children, will develop coronary artery disease. As a multifactorial condition, lifestyle factors also contribute to your risk of developing coronary artery disease.

This test is not intended to diagnose a disease or tell you anything about your risk for developing a disease in the future. Talk with your health care provider for more information if you are concerned about your risk of coronary artery disease.

A genetic consultation may be of benefit. Please contact the independent genetic counseling provider who reviewed your test order and results, PWNHealth toll free at 1-888-494-7333 or by email at gc@pwnhealth.com, if you have questions regarding this report.

Method: The variants interpreted by the Mayo Clinic GeneGuide™ system are detected by our partner, Helix. The Helix Exome+ assay uses next generation sequencing (NGS) of DNA to analyze the entire human exome plus several hundred thousand additional informative regions outside of the exome, including the complete mitochondrial genome. The Helix Exome+ assay was performed on genomic DNA extracted from your submitted saliva sample. DNA from your sample was captured and enriched using a custom set of reagents for target selection. Next Generation Sequencing was performed on an Illumina DNA sequencing instrument using HapMap samples NA12878 and NA12877 as internal controls. Alignment to a modified version of GRCh38 and variant calling were completed using a customized version of Sentieon's DNAseq software. The Helix bioinformatics pipeline applied several quality control filters to produce the final passing variants list. Using a semi-automated platform for laboratory review and reporting, The Mayo Clinic GeneGuide™ interpretive system then pulled a predetermined set of clinically validated variants from the Helix Secure Database, and matched the variants to the appropriate result interpretation.

Variants Tested: The variant nomenclature that is used to describe the Coronary Artery Disease variants included in Mayo Clinic GeneGuide™ is based on:

NM_001408.2(SORT1)

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

c.*1859C>T

NM_005577.2(LPA)

c.5673A>G (p.I1891M)

NC_000009.12

g.22098575A>G

Cautions:

- The interpretation of this report is based upon the information provided by the ordering physician at the time of testing. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.
- This test is designed to detect the specific clinically validated variants listed in this report. It is possible that other types of disease-causing variants exist in this or other related genes that were not detected.
- Patients who have ever had an allogeneic blood or marrow transplant or who have received a heterologous blood transfusion within the last month, can have inaccurate genetic test results due to presence of donor DNA. Patients who have undergone liver transplantation may also have inaccurate genetic test results.
- Variant nomenclature is based on genomic build GRCh38. Variants are evaluated and classified according to ACMG recommendations (Richards, et al., 2015). Variant classification may change over time. If a variant that was part of Mayo Clinic GeneGuide™ is reclassified and you have questions, please contact us at 1-877-858-0398.

Cautions for Health Care Providers:

- This test is not intended to diagnose a disease, determine medical treatment, or tell the user anything about their current state of health.
- This test is intended to provide users with their genetic information to inform lifestyle decisions and conversations with their doctor or other healthcare professional.
- Any diagnostic or treatment decisions should be based on testing and/or other information that your healthcare provider determines to be appropriate for you.

Warnings:

- This test provides genetic risk information based on assessment of specific genetic variants but does not report on a user's entire genetic profile.
- This test does not detect all genetic variants related to a given disease, and the absence of a variant tested does not rule out the presence of other genetic variants that may be related to the disease
- Other companies offering a genetic risk test may be detecting different genetic variants for the same disease, so the user may get different results using a test from a different company.
- Factors such as environmental and lifestyle risk factors may affect the risk of developing a given disease.
- Some people may feel anxious about getting genetic test health results. This is normal. If the potential user feels very anxious, such user should speak to his or her doctor or other healthcare professional prior to collection of a sample for testing. This test is not a substitute for visits to a doctor or other healthcare professional. Users should consult with their doctor or other healthcare professional if they have any questions or concerns about the results of their test or their current state of health.
- This test is not intended to diagnose a disease, tell you anything about your current state of health, or be used to make medical decisions, including whether or not you should take a medication or how much of a medication you should take.

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Venous Thromboembolism Results

Condition	Gene	# Variants	Variant(s) Identified	Variant(s) Classification	Interpretation
Venous Thromboembolism	F5	1	c.1601G>A (p.Arg534Gln)	Risk allele	Increased risk
	F2	0			

Venous Thromboembolism interpretation

This test found one variant in the F5 (factor V) gene in your DNA. In people with one factor V variant, the risk of having a harmful blood clot is around 3 to 5 times higher than in people without a variant. The vast majority of people with one factor V variant never develop harmful blood clots. The risk of developing harmful blood clots increases with age. Certain lifestyle choices can make the risk of harmful blood clots higher in someone with one factor V variant.

Venous thromboembolism (VTE) is a condition in which one or more abnormal blood clots form in a vein. These clots can cause medical complications and may even be life-threatening. In VTE, blood clots form due to changes in blood flow, damage to blood vessels and risk factors for abnormal clotting. VTE includes two conditions, deep vein thrombosis (DVT) and pulmonary embolism (PE). VTE can arise from genetic risk factors, called inherited thrombophilia. Approximately 5 to 8 percent of the U.S. population has a gene variant that causes inherited thrombophilia. Inherited thrombophilia is an autosomal dominant condition, so inheriting a gene variant from only one parent increases VTE risk. Having two variants (one from each parent) causes a higher risk of VTE. The two most common gene variants are in the genes F5 (called coagulation factor V) on chromosome 1 and F2 (called coagulation factor II) on chromosome 11. Mayo Clinic GeneGuide™ tests for the most common variant in each gene. Many people who inherit these gene variants never develop VTE. Nongenetic risk factors such as age, hormones, obesity, smoking or lack of movement for a long period of time also can cause VTE. Share these results with your health care provider who can discuss how this result impacts your health. Consider talking with your family members about their chances of having a genetic variant in F5, too. Discussion guides for talking with your family and health care provider are available in the Mayo Clinic GeneGuide™ app.

This test is not intended to diagnose a disease or tell you anything about your risk for developing a disease in the future.

A genetic consultation may be of benefit. Please contact the independent genetic counseling provider who reviewed your test order and results, PWNHealth toll free at 1-888-494-7333 or by email at gc@pwnhealth.com, if you have questions regarding this report.

Method: The variants interpreted by the Mayo Clinic GeneGuide™ system are detected by our partner, Helix. The Helix Exome+ assay uses next generation sequencing (NGS) of DNA to analyze the entire human exome plus several hundred thousand additional informative regions outside of the exome, including the complete mitochondrial genome. The Helix Exome+ assay was performed on genomic DNA extracted from your submitted saliva sample. DNA from your sample was captured and enriched using a custom set of reagents for target selection. Next Generation Sequencing was performed on an Illumina DNA sequencing instrument using HapMap samples NA12878 and NA12877 as internal controls. Alignment to a modified version of GRCh38 and variant calling were completed using a customized version of Sentieon's DNAseq software. The Helix bioinformatics pipeline applied several quality control filters to produce the final passing variants list. Using a semi-automated platform for laboratory review and reporting, The Mayo Clinic GeneGuide™ interpretive system then pulled a predetermined set of clinically validated variants from the Helix Secure Database, and matched the variants to the appropriate result interpretation.

Variants Tested: The variant nomenclature that is used to describe the Venous Thromboembolism variants included in Mayo Clinic GeneGuide™ is based on:

NM_000130.4(F5)
c.1601G>A (p.Arg534Gln)

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

NM_000506.4(F2)
c.*97G>A

Cautions:

- The interpretation of this report is based upon the information provided by the ordering physician at the time of testing. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.
- This test is designed to detect the specific clinically validated variants listed in this report. It is possible that other types of disease-causing variants exist in this or other related genes that were not detected.
- Patients who have ever had an allogeneic blood or marrow transplant or who have received a heterologous blood transfusion within the last month, can have inaccurate genetic test results due to presence of donor DNA. Patients who have undergone liver transplantation may also have inaccurate genetic test results.
- Variant nomenclature is based on genomic build GRCh38. Variants are evaluated and classified according to ACMG recommendations (Richards, et al., 2015). Variant classification may change over time. If a variant that was part of Mayo Clinic GeneGuide™ is reclassified and you have questions, please contact us at 1-877-858-0398.

Cautions for Health Care Providers:

- This test is not intended to diagnose a disease, determine medical treatment, or tell the user anything about their current state of health.
- This test is intended to provide users with their genetic information to inform lifestyle decisions and conversations with their doctor or other healthcare professional.
- Any diagnostic or treatment decisions should be based on testing and/or other information that your healthcare provider determines to be appropriate for you.

Warnings:

- This test provides genetic risk information based on assessment of specific genetic variants but does not report on a user's entire genetic profile.
- This test does not detect all genetic variants related to a given disease, and the absence of a variant tested does not rule out the presence of other genetic variants that may be related to the disease
- Other companies offering a genetic risk test may be detecting different genetic variants for the same disease, so the user may get different results using a test from a different company.
- Factors such as environmental and lifestyle risk factors may affect the risk of developing a given disease.
- Some people may feel anxious about getting genetic test health results. This is normal. If the potential user feels very anxious, such user should speak to his or her doctor or other healthcare professional prior to collection of a sample for testing. This test is not a substitute for visits to a doctor or other healthcare professional. Users should consult with their doctor or other healthcare professional if they have any questions or concerns about the results of their test or their current state of health.
- This test is not intended to diagnose a disease, tell you anything about your current state of health, or be used to make medical decisions, including whether or not you should take a medication or how much of a medication you should take.

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Introduction to Health Traits

When you look at the individuals in a random group of 100 adults who aren't related, you will notice an amazing variety of physical features. Differences in faces, height, weight, skin color and hair thickness may be some of the variations you notice first. To varying degrees, these traits are determined by genes. Given that no two people are exactly alike, it's amazing to realize that individual human genomes are identical at 999 out of 1000 places.

Besides affecting physical appearance, genetic variants are also responsible for a great deal of variety in specific body functions, such as digestion, metabolism and immune system reactions. Mayo Clinic GeneGuide™ classifies these genetic variants as health traits. Hundreds of traits have been identified that could fall into this category — high sensitivity to bitter flavors, for example, or exaggerated physical responses to stressful but temporary situations.

Most health traits seem to have complex or multifactorial inheritance, meaning that they are the result of interactions among multiple genes and nongenetic factors. A few, like blood types, can be passed to children in predictable patterns.

Some health traits are indicators of an increased risk for a more serious disease with certain lifestyles. Therefore, knowing you have certain health traits can help you make lifestyle choices that will help you feel your best all the time.

Health Traits Result Summary

Condition	Gene	# Variants	Variant(s) Identified	Variant(s) Classification	Interpretation
Atopic Dermatitis	FLG	0			No variants identified
Alcohol Flush Reaction	ALDH2	0			No variants identified
Lactase Persistence	MCM6	2	c.1917+326C>T	Risk allele	Lactase persistent
		2	c.1362+117G>A	Risk allele	

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Atopic Dermatitis Results

Condition	Gene	# Variants	Variant(s) Identified	Variant(s) Classification	Interpretation
Atopic Dermatitis	FLG	0			No variants identified

Atopic Dermatitis interpretation

This test did not identify variants in the FLG gene associated with an increased risk of atopic dermatitis or ichthyosis vulgaris. Even though this test didn't identify any variants, there is still a chance that you could have a different variant in FLG or other genes associated with atopic dermatitis or ichthyosis vulgaris. If you have a personal or family history of atopic dermatitis, you may have other factors that increase your risk.

Atopic dermatitis (eczema) is a condition that causes the skin to become red, itchy and prone to rashes due to a problem with the skin's outer protective barrier. In some people with atopic dermatitis, the skin becomes sensitive to irritants that can cause allergic reactions. Atopic dermatitis may be accompanied by asthma or hay fever. Atopic dermatitis most commonly occurs in childhood, often before the age of 5. It typically improves with age, but an affected person may be more likely to develop contact allergies to things such as fragrance, nickel or adhesive tapes. Less commonly, atopic dermatitis is long lasting (chronic). In these cases, signs and symptoms tend to come and go, with occasional short-term episodes of worsening (flares). A shortage or lack of filaggrin protein also interferes with the ability of the skin to retain moisture, which can lead to dry skin (as in ichthyosis vulgaris). Atopic dermatitis is a multifactorial condition. This means that genetic and nongenetic factors play a role in its development. One genetic factor that is associated with the development of atopic dermatitis is having a variant the filaggrin (FLG) gene on chromosome 1. FLG instructs cells to make filaggrin protein, which helps to create the tough outermost layer of skin. However, not all people with atopic dermatitis have a variant in FLG, and not all people with an FLG variant will have atopic dermatitis. People with one variant have a 50 percent chance of passing on the variant to each of their children. People with two variants always pass on a variant to each of their children.

This test is not intended to diagnose a disease or tell you anything about your risk for developing a disease in the future.

For most people, there's nothing more to do based on this result. If you have concerns about an ongoing skin condition, talk with your health care provider.

A genetic consultation may be of benefit. Please contact the independent genetic counseling provider who reviewed your test order and results, PWNHealth toll free at 1-888-494-7333 or by email at gc@pwnhealth.com, if you have questions regarding this report.

Method: The variants interpreted by the Mayo Clinic GeneGuide™ system are detected by our partner, Helix. The Helix Exome+ assay uses next generation sequencing (NGS) of DNA to analyze the entire human exome plus several hundred thousand additional informative regions outside of the exome, including the complete mitochondrial genome. The Helix Exome+ assay was performed on genomic DNA extracted from your submitted saliva sample. DNA from your sample was captured and enriched using a custom set of reagents for target selection. Next Generation Sequencing was performed on an Illumina DNA sequencing instrument using HapMap samples NA12878 and NA12877 as internal controls. Alignment to a modified version of GRCh38 and variant calling were completed using a customized version of Sentieon's DNAseq software. The Helix bioinformatics pipeline applied several quality control filters to produce the final passing variants list. Using a semi-automated platform for laboratory review and reporting, The Mayo Clinic GeneGuide™ interpretive system then pulled a predetermined set of clinically validated variants from the Helix Secure Database, and matched the variants to the appropriate result interpretation.

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Variants Tested: The variant nomenclature that is used to describe the Atopic Dermatitis variants included in Mayo Clinic GeneGuide™ is based on:

NM_002016.1(FLG)

c.2282_2285delCAGT (p.Ser761Cysfs), c.1501C>T (p.Arg501Ter)

Cautions:

- The interpretation of this report is based upon the information provided by the ordering physician at the time of testing. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.
- This test is designed to detect the specific clinically validated variants listed in this report. It is possible that other types of disease-causing variants exist in this or other related genes that were not detected.
- Patients who have ever had an allogeneic blood or marrow transplant or who have received a heterologous blood transfusion within the last month, can have inaccurate genetic test results due to presence of donor DNA. Patients who have undergone liver transplantation may also have inaccurate genetic test results.
- Variant nomenclature is based on genomic build GRCh38. Variants are evaluated and classified according to ACMG recommendations (Richards, et al., 2015). Variant classification may change over time. If a variant that was part of Mayo Clinic GeneGuide™ is reclassified and you have questions, please contact us at 1-877-858-0398.

Cautions for Health Care Providers:

- This test is not intended to diagnose a disease, determine medical treatment, or tell the user anything about their current state of health.
- This test is intended to provide users with their genetic information to inform lifestyle decisions and conversations with their doctor or other healthcare professional.
- Any diagnostic or treatment decisions should be based on testing and/or other information that your healthcare provider determines to be appropriate for you.

Warnings:

- This test provides genetic risk information based on assessment of specific genetic variants but does not report on a user's entire genetic profile.
- This test does not detect all genetic variants related to a given disease, and the absence of a variant tested does not rule out the presence of other genetic variants that may be related to the disease
- Other companies offering a genetic risk test may be detecting different genetic variants for the same disease, so the user may get different results using a test from a different company.
- Factors such as environmental and lifestyle risk factors may affect the risk of developing a given disease.
- Some people may feel anxious about getting genetic test health results. This is normal. If the potential user feels very anxious, such user should speak to his or her doctor or other healthcare professional prior to collection of a sample for testing. This test is not a substitute for visits to a doctor or other healthcare professional. Users should consult with their doctor or other healthcare professional if they have any questions or concerns about the results of their test or their current state of health.
- This test is not intended to diagnose a disease, tell you anything about your current state of health, or be used to make medical decisions, including whether or not you should take a medication or how much of a medication you should take.

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Alcohol Flush Reaction Results

Condition	Gene	# Variants	Variant(s) Identified	Variant(s) Classification	Interpretation
Alcohol Flush Reaction	ALDH2	0			No variants identified

Alcohol Flush Reaction interpretation

This test did not identify variants in the ALDH2 gene associated with alcohol flush reaction. You are unlikely to have inherited alcohol flush reaction. Even though this test didn't identify any variants, there is a chance that you still could have a different genetic variant this test was not designed to detect. This test is not designed to identify other causes associated with alcohol flush reaction.

Alcohol flush reaction is a genetic trait that interferes with the body's ability to break down alcohol. When people with this trait drink alcohol, they experience facial redness (flushing), rapid heartbeat, nausea, and low blood pressure. This happens because one of the several enzymes that normally break down alcohol (ALDH2) does not function properly. This causes a substance called acetaldehyde (a-set-aldi-hide) to build up in the body. When acetaldehyde builds up, people can develop the signs and symptoms of alcohol flush. In addition to causing alcohol flush, people with the trait have an increased risk of cancer of the esophagus (the connection between your mouth and stomach), and other health risks, if they regularly drink alcohol. Alcohol flush reaction is an inherited trait caused by variants in the ALDH2 gene located on chromosome 12. The most common variant in ALDH2, tested by Mayo Clinic GeneGuide™, is rs671. This variant is inherited in an autosomal dominant fashion, so having one or more variants in rs671 causes alcohol flush reaction. Inheriting two variants (one from each parent) causes more severe reaction. People with one variant have a 50 percent chance of passing on the variant to each of their children. People with two variants always pass on a variant to each of their children.

This test is not intended to diagnose a disease or tell you anything about your risk for developing a disease in the future. If you need help to stop drinking alcohol, there are treatment programs available to help you. Learn more at http://www.mayoclinic.org/diseases-conditions/drugaddiction/basics/treatment/con-20020970.

A genetic consultation may be of benefit. Please contact the independent genetic counseling provider who reviewed your test order and results, PWNHealth toll free at 1-888-494-7333 or by email at gc@pwnhealth.com, if you have questions regarding this report.

Method: The variants interpreted by the Mayo Clinic GeneGuide™ system are detected by our partner, Helix. The Helix Exome+ assay uses next generation sequencing (NGS) of DNA to analyze the entire human exome plus several hundred thousand additional informative regions outside of the exome, including the complete mitochondrial genome. The Helix Exome+ assay was performed on genomic DNA extracted from your submitted saliva sample. DNA from your sample was captured and enriched using a custom set of reagents for target selection. Next Generation Sequencing was performed on an Illumina DNA sequencing instrument using HapMap samples NA12878 and NA12877 as internal controls. Alignment to a modified version of GRCh38 and variant calling were completed using a customized version of Sentieon's DNAseq software. The Helix bioinformatics pipeline applied several quality control filters to produce the final passing variants list. Using a semi-automated platform for laboratory review and reporting, The Mayo Clinic GeneGuide™ interpretive system then pulled a predetermined set of clinically validated variants from the Helix Secure Database, and matched the variants to the appropriate result interpretation.

Variants Tested: The variant nomenclature that is used to describe the Alcohol Flush Reaction variants included in Mayo Clinic GeneGuide™ is based on:

NM_000690.3(ALDH2)
c.1510G>A (p.Glu504Lys)

Cautions:

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

- The interpretation of this report is based upon the information provided by the ordering physician at the time of testing. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.
- This test is designed to detect the specific clinically validated variants listed in this report. It is possible that other types of disease-causing variants exist in this or other related genes that were not detected.
- Patients who have ever had an allogeneic blood or marrow transplant or who have received a heterologous blood transfusion within the last month, can have inaccurate genetic test results due to presence of donor DNA. Patients who have undergone liver transplantation may also have inaccurate genetic test results.
- Variant nomenclature is based on genomic build GRCh38. Variants are evaluated and classified according to ACMG recommendations (Richards, et al., 2015). Variant classification may change over time. If a variant that was part of Mayo Clinic GeneGuide™ is reclassified and you have questions, please contact us at 1-877-858-0398.

Cautions for Health Care Providers:

- This test is not intended to diagnose a disease, determine medical treatment, or tell the user anything about their current state of health.
- This test is intended to provide users with their genetic information to inform lifestyle decisions and conversations with their doctor or other healthcare professional.
- Any diagnostic or treatment decisions should be based on testing and/or other information that your healthcare provider determines to be appropriate for you.

Warnings:

- This test provides genetic risk information based on assessment of specific genetic variants but does not report on a user's entire genetic profile.
- This test does not detect all genetic variants related to a given disease, and the absence of a variant tested does not rule out the presence of other genetic variants that may be related to the disease
- Other companies offering a genetic risk test may be detecting different genetic variants for the same disease, so the user may get different results using a test from a different company.
- Factors such as environmental and lifestyle risk factors may affect the risk of developing a given disease.
- Some people may feel anxious about getting genetic test health results. This is normal. If the potential user feels very anxious, such user should speak to his or her doctor or other healthcare professional prior to collection of a sample for testing. This test is not a substitute for visits to a doctor or other healthcare professional. Users should consult with their doctor or other healthcare professional if they have any questions or concerns about the results of their test or their current state of health.
- This test is not intended to diagnose a disease, tell you anything about your current state of health, or be used to make medical decisions, including whether or not you should take a medication or how much of a medication you should take.

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

Lactase Persistence Results

Condition	Gene	# Variants	Variant(s) Identified	Variant(s) Classification	Interpretation
Lactase Persistence	MCM6	2	c.1917+326C>T	Risk allele	Lactase persistent
		2	c.1362+117G>A	Risk allele	

Lactase Persistence interpretation

This test found two or more genetic variants in the MCM6 gene. People with one or more genetic variants in MCM6 usually have lactase persistence. You likely do not have the signs and symptoms of lactose intolerance.

Lactase persistence is a genetic trait that affects the body's ability to digest dairy products. Lactose (with an "o") is a sugar found in human, cow and goat milk. Lactase (with an "a") is an enzyme that helps digest lactose. People with low levels of lactase are lactase non-persistent. They have difficulty digesting dairy products, also known as lactose intolerance, beyond childhood. Symptoms of lactose intolerance can include bloating and abdominal pain, nausea, gas and diarrhea. However, people with lactase persistence can easily digest milk products throughout their lifetime. Lactase persistence is an inherited trait caused by variants in the MCM6 gene located on chromosome 2.

Lactase persistence is passed down from parents to children in an autosomal dominant pattern, so having one or more variants in the MCM6 genes leads to lactase persistence. The risk of passing on a MSM6 variant depends on how many variants you have. People with one variant have a 50 percent chance of passing on the variant to each of their children. When a person has two or more MSM6 variants, the risk of passing on at least one MSMG variant that causes lactase persistence to each of their children ranges from 50-100 percent. This test is not intended to diagnose a disease or tell you anything about your risk for developing a disease in the future.

A genetic consultation may be of benefit. Please contact the independent genetic counseling provider who reviewed your test order and results, PWNHealth toll free at 1-888-494-7333 or by email at gc@pwnhealth.com, if you have questions regarding this report.

Method: The variants interpreted by the Mayo Clinic GeneGuide™ system are detected by our partner, Helix. The Helix Exome+ assay uses next generation sequencing (NGS) of DNA to analyze the entire human exome plus several hundred thousand additional informative regions outside of the exome, including the complete mitochondrial genome. The Helix Exome+ assay was performed on genomic DNA extracted from your submitted saliva sample. DNA from your sample was captured and enriched using a custom set of reagents for target selection. Next Generation Sequencing was performed on an Illumina DNA sequencing instrument using HapMap samples NA12878 and NA12877 as internal controls. Alignment to a modified version of GRCh38 and variant calling were completed using a customized version of Sentieon's DNAseq software. The Helix bioinformatics pipeline applied several quality control filters to produce the final passing variants list. Using a semi-automated platform for laboratory review and reporting, The Mayo Clinic GeneGuide™ interpretive system then pulled a predetermined set of clinically validated variants from the Helix Secure Database, and matched the variants to the appropriate result interpretation.

Variants Tested: The variant nomenclature that is used to describe the Lactase Persistence variants included in Mayo Clinic GeneGuide™ is based on:

NM_005915.5(MCM6)

c.1917+326C>T, c.1362+117G>A

Cautions:

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.

- The interpretation of this report is based upon the information provided by the ordering physician at the time of testing. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.
- This test is designed to detect the specific clinically validated variants listed in this report. It is possible that other types of disease-causing variants exist in this or other related genes that were not detected.
- Patients who have ever had an allogeneic blood or marrow transplant or who have received a heterologous blood transfusion within the last month, can have inaccurate genetic test results due to presence of donor DNA. Patients who have undergone liver transplantation may also have inaccurate genetic test results.
- Variant nomenclature is based on genomic build GRCh38. Variants are evaluated and classified according to ACMG recommendations (Richards, et al., 2015). Variant classification may change over time. If a variant that was part of Mayo Clinic GeneGuide™ is reclassified and you have questions, please contact us at 1-877-858-0398.

Cautions for Health Care Providers:

- This test is not intended to diagnose a disease, determine medical treatment, or tell the user anything about their current state of health.
- This test is intended to provide users with their genetic information to inform lifestyle decisions and conversations with their doctor or other healthcare professional.
- Any diagnostic or treatment decisions should be based on testing and/or other information that your healthcare provider determines to be appropriate for you.

Warnings:

- This test provides genetic risk information based on assessment of specific genetic variants but does not report on a user's entire genetic profile.
- This test does not detect all genetic variants related to a given disease, and the absence of a variant tested does not rule out the presence of other genetic variants that may be related to the disease
- Other companies offering a genetic risk test may be detecting different genetic variants for the same disease, so the user may get different results using a test from a different company.
- Factors such as environmental and lifestyle risk factors may affect the risk of developing a given disease.
- Some people may feel anxious about getting genetic test health results. This is normal. If the potential user feels very anxious, such user should speak to his or her doctor or other healthcare professional prior to collection of a sample for testing. This test is not a substitute for visits to a doctor or other healthcare professional. Users should consult with their doctor or other healthcare professional if they have any questions or concerns about the results of their test or their current state of health.
- This test is not intended to diagnose a disease, tell you anything about your current state of health, or be used to make medical decisions, including whether or not you should take a medication or how much of a medication you should take.

Laboratory Testing Facilities

Interpretive report performed and validated by Mayo Clinic GeneGuide™ (MCGG) Laboratory; 200 First St SW Rochester, MN 55905. NGS performed and validated by Helix 9875 Towne Center Dr., San Diego, CA 92121. This test has not been cleared or approved by the U.S. Food and Drug Administration.