Patient Information	Specimen Information	Client Information
WARDEN, CHARLES **DOB: 04/05/1985 AGE: 38** Gender: M Phone: 404.316.0012 Patient ID: 20220711230258006735	Specimen: GS006771A Requisition: 1221569 Lab Ref #: GBFIWJWQ547 Collected: 11/15/2023 Received: 11/22/2023 / 16:47 EDT Reported: 10/30/2024 / 13:49 EDT	Client #: 97554902 MAIL992 MOTA, CHRISTINA QD CIT-ECOMMERCE 500 PLAZA DR SECAUCUS, NJ 07094-3619

Genetic Insights Health Screening Test

Abnormal Reclassfication Uninformative Amended

Heart and Blood Health

	Condition	Gene(s)
	Cardiomyopathy	MYBPC3, MYH7
	Familial Hypercholesterolemia	APOB, LDLR, PCSK9
	Hereditary Hemochromatosis	HFE
⚠	Hereditary Thrombophilia	F2, F5

Carrier Status

	Condition	Gene(s)
⚠	Cystic Fibrosis	CFTR
	Sickle Cell Anemia	HBB
	Tay-Sachs Disease	HEXA

Patient Information	Specimen Information	Client Information
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Genetic Insights Health Screening Test

Abnormal **Reclassfication** **Uninformative** **Amended**

Cancer Risk

Condition	Gene(s)
APC-Associated Hereditary Cancer	APC
ATM-Associated Hereditary Cancer	ATM
CHEK2-Associated Hereditary Cancer	CHEK2
Hereditary Breast and Ovarian Cancer	BRCA1, BRCA2
Juvenile Polyposis Syndrome (BMPR1A-associated)	BMPR1A
Juvenile Polyposis Syndrome / Hereditary Hemorrhagic Telangiectasia (SMAD4-associated)	SMAD4
Lynch Syndrome	MLH1, MSH2, MSH6, PMS2
MUTYH-Associated Polyposis	MUTYH
PALB2-Associated Hereditary Cancer	PALB2
Peutz-Jeghers Syndrome	STK11
POLD1/POLE-Associated Hereditary Cancer	POLD1, POLE

Connective Tissue Disorders

Condition	Gene(s)
Classical Ehlers-Danlos Syndrome	COL5A1, COL5A2
Familial Thoracic Aortic Aneurysm and Dissection	ACTA2
Loeys-Dietz Syndrome	TGFB2, TGFB3, TGFBR1, TGFBR2
Marfan Syndrome	FBN1
Vascular Ehlers-Danlos Syndrome	COL3A1

Patient Information	Specimen Information	Client Information
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Genetic Insights Health Screening Test

Healthcare Provider Quick Reference Guides

Let us make it easy for you to know how best to support your patients who have genetic variants.

Scan a QR code for your Quick Reference Guide with:

* Next steps and clinical guidelines
* Conversation starters
* Condition details

Hereditary Thrombophilia Quick Reference Guide

Cystic Fibrosis Quick Reference Guide

Report Id : FAA47DE6A3E389D3159B1DA8A7ED0CD49C471EDF649D9AC7C3CBD42C463959B6

Patient Information	Specimen Information	Client Information
WARDEN, CHARLES **DOB: 04/05/1985 AGE: 38** Gender: M Patient ID: 20220711230258006735	Specimen: GS006771A Collected: 11/15/2023 Received: 11/22/2023 / 16:47 EDT Reported: 10/30/2024 / 13:49 EDT	Client #: 97554902 MOTA, CHRISTINA

Cardiomyopathy

Result

NO CLINICALLY RELEVANT VARIANTS DETECTED

Clinical Relevance

No clinically relevant variants associated with an increased risk of cardiomyopathy were detected in the MYBPC3 or MYH7 genes. As a screening test, this assay is not intended to rule out all variants in the gene(s) tested.

Tested Genes Include:
MYBPC3, MYH7

Reviewer

Vivekananda Datta, MD, PhD

Methods and Limitations

This screening test should not be used for diagnosis without confirmation by other medically established means. When a clinically relevant variant(s) is identified, confirmatory testing is strongly recommended prior to making any clinical decisions. A negative result from this analysis does not rule out the possibility that the tested individual carries a rare unexamined variant, variant in an undetectable region, or a type of variant not intended to be detected by the test. For complete methods and limitations, please refer to the end of this report. If applicable, condition specific additional information can be found below.

Condition Specific Additional Information

The intended use of this screening test is to identify pathogenic and likely pathogenic variants in the listed genes associated with an increased risk of developing cardiomyopathy, which may include, but is not limited to, hypertrophic cardiomyopathy, dilated cardiomyopathy, and left ventricular non-compaction, either alone or as part of a distal myopathy phenotype.

Healthcare Provider Resources

For general information about this test, visit www.questdiagnostics.com/genetic-health-screening

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WARDEN, CHARLES **DOB: 04/05/1985 AGE: 38** Gender: M Patient ID: 20220711230258006735	Specimen: GS006771A Collected: 11/15/2023 Received: 11/22/2023 / 16:47 EDT Reported: 10/30/2024 / 13:49 EDT	Client #: 97554902 MOTA, CHRISTINA

Familial Hypercholesterolemia

Result

NO CLINICALLY RELEVANT VARIANTS DETECTED

Clinical Relevance

No clinically relevant variants associated with familial hypercholesterolemia were detected in the APOB, LDLR or PCSK9 genes. As a screening test, this assay is not intended to rule out all variants in the gene(s) tested.

Tested Genes Include:
APOB, LDLR, PCSK9

Reviewer

Vivekananda Datta, MD, PhD

Methods and Limitations

This screening test should not be used for diagnosis without confirmation by other medically established means. When a clinically relevant variant(s) is identified, confirmatory testing is strongly recommended prior to making any clinical decisions. A negative result from this analysis does not rule out the possibility that the tested individual carries a rare unexamined variant, variant in an undetectable region, or a type of variant not intended to be detected by the test. For complete methods and limitations, please refer to the end of this report. If applicable, condition specific additional information can be found below.

Healthcare Provider Resources

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WARDEN, CHARLES **DOB: 04/05/1985 AGE: 38** Gender: M Patient ID: 20220711230258006735	Specimen: GS006771A Collected: 11/15/2023 Received: 11/22/2023 / 16:47 EDT Reported: 10/30/2024 / 13:49 EDT	Client #: 97554902 MOTA, CHRISTINA

Hereditary Hemochromatosis

Result

NO CLINICALLY RELEVANT VARIANTS DETECTED

Clinical Relevance

The selected variant(s) associated with hereditary hemochromatosis were not detected in the HFE gene.

Results

Gene	Variant	Interpretation	Zygosity
HFE	c.187C>G (p.His63Asp)	NOT DETECTED	NOT DETECTED
HFE	c.845G>A (p.Cys282Tyr)	NOT DETECTED	NOT DETECTED

Tested Genes Include:
HFE (selected variants only: c.187C>G, c.845G>A)

Reviewer

Vivekananda Datta, MD, PhD

Methods and Limitations

This screening test should not be used for diagnosis without confirmation by other medically established means. When a clinically relevant variant(s) is identified, confirmatory testing is strongly recommended prior to making any clinical decisions. A negative result from this analysis does not rule out the possibility that the tested individual carries a rare unexamined variant, variant in an undetectable region, or a type of variant not intended to be detected by the test. For complete methods and limitations, please refer to the end of this report. If applicable, condition specific additional information can be found below.

Healthcare Provider Resources

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Hereditary Thrombophilia

Result

CLINICALLY RELEVANT VARIANT(S) DETECTED

Scan for your Quick Reference Guide

Clinical Relevance

A pathogenic variant was detected in the F5 gene. The presence of a single pathogenic variant in the F5 gene is associated with hereditary thrombophilia.

Results

Gene	Variant	Interpretation	Zygosity
F5	c.1601G>A (p.Arg534Gln)	PATHOGENIC	Heterozygous

Tested Genes Include:
F2 (selected variants only: c.*97G>A), F5 (selected variants only: c.1601G>A)

F5 c.1601G>A (p.Arg534Gln) Variant Details

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

- This variant is expected to change a single amino acid to a different amino acid at the same position within the protein.

- Published literature regarding this variant is consistent with a pathogenic classification (PMID: 8164741, 7911872, 7910348, 23900608, 21116184, 16024978).

- The frequency of this variant in the general population is consistent with a pathogenic classification (http://gnomad.broadinstitute.org).

- ClinVar contains an entry for this variant: 642

Reviewer

Ian M. Wilson, PhD, FACMG [SJC21]

Methods and Limitations

This screening test should not be used for diagnosis without confirmation by other medically established means. When a clinically relevant variant(s) is identified, confirmatory testing is strongly recommended prior to making any clinical decisions. A negative result from this analysis does not rule out the possibility that the tested individual carries a rare unexamined variant, variant in an undetectable region, or a type of variant not intended to be detected by the test. For complete methods and limitations, please refer to the end of this report. If applicable, condition specific additional information can be found below.

Healthcare Provider Resources

Let us make it easy for you to know how best to support your patients who have genetic variants. Find your Quick Reference Guide with next steps, clinical guidelines, conversation starters, and condition details at www.questdiagnostics.com/genetic-health-screening/F5_1_variant.pdf, or scan the QR code above.

Patient Information	Specimen Information	Client Information
WARDEN, CHARLES **DOB: 04/05/1985 AGE: 38** Gender: M Patient ID: 20220711230258006735	Specimen: GS006771A Collected: 11/15/2023 Received: 11/22/2023 / 16:47 EDT Reported: 10/30/2024 / 13:49 EDT	Client #: 97554902 MOTA, CHRISTINA

Cystic Fibrosis

Result

CLINICALLY RELEVANT VARIANT(S) DETECTED

Scan for your Quick Reference Guide

Clinical Relevance

A pathogenic variant was detected in the CFTR gene. The presence of a single pathogenic variant in the CFTR gene is associated with a carrier status for cystic fibrosis.

Results

Gene	Variant	Interpretation	Zygosity
CFTR	c.262_263del (p.Leu88Ilefs*22)	PATHOGENIC	Heterozygous

Tested Genes Include:
CFTR

*CFTR c.262_263del (p.Leu88Ilefs*22)* Variant Details

CFTR c.262_263del (p.Leu88Ilefs*22) (NM_000492.3):

- This variant is expected to cause premature truncation of the protein.

- Published literature regarding this variant is consistent with a pathogenic classification (PMID: 7691344, 7509310, 32429104, 22658665, 18456578, 16051530).

- The frequency of this variant in the general population is consistent with a pathogenic classification (http://gnomad.broadinstitute.org).

- ClinVar contains an entry for this variant: 7232

Reviewer

Ian M. Wilson, PhD, FACMG [SJC21]

Methods and Limitations

This screening test should not be used for diagnosis without confirmation by other medically established means. When a clinically relevant variant(s) is identified, confirmatory testing is strongly recommended prior to making any clinical decisions. A negative result from this analysis does not rule out the possibility that the tested individual carries a rare unexamined variant, variant in an undetectable region, or a type of variant not intended to be detected by the test. For complete methods and limitations, please refer to the end of this report. If applicable, condition specific additional information can be found below.

Condition Specific Additional Information

This assay excludes detection of the presence or absence of the R117H variant and does not genotype the intron 8 poly-T tract. This assay excludes detection of DNA variants associated only with congenital bilateral absence of the vas deferens.

Healthcare Provider Resources

Let us make it easy for you to know how best to support your patients who have genetic variants. Find your Quick Reference Guide with next steps, clinical guidelines, conversation starters, and condition details at www.questdiagnostics.com/genetic-health-screening/CF_carrier.pdf, or scan the QR code above.

Patient Information	Specimen Information	Client Information
WARDEN, CHARLES **DOB: 04/05/1985 AGE: 38** Gender: M Patient ID: 20220711230258006735	Specimen: GS006771A Collected: 11/15/2023 Received: 11/22/2023 / 16:47 EDT Reported: 10/30/2024 / 13:49 EDT	Client #: 97554902 MOTA, CHRISTINA

Sickle Cell Anemia

Result

NO CLINICALLY RELEVANT VARIANTS DETECTED

Clinical Relevance

The selected variant(s) associated with sickle cell anemia were not detected in the HBB gene.

Results

Gene	Variant	Interpretation	Zygosity
HBB	c.20A>T (p.Glu7Val)	NOT DETECTED	NOT DETECTED

Tested Genes Include:
HBB (selected variants only: c.20A>T)

Reviewer

Vivekananda Datta, MD, PhD

Methods and Limitations

This screening test should not be used for diagnosis without confirmation by other medically established means. When a clinically relevant variant(s) is identified, confirmatory testing is strongly recommended prior to making any clinical decisions. A negative result from this analysis does not rule out the possibility that the tested individual carries a rare unexamined variant, variant in an undetectable region, or a type of variant not intended to be detected by the test. For complete methods and limitations, please refer to the end of this report. If applicable, condition specific additional information can be found below.

Healthcare Provider Resources

For general information about this test, visit www.questdiagnostics.com/genetic-health-screening

Patient Information	Specimen Information	Client Information
WARDEN, CHARLES **DOB: 04/05/1985 AGE: 38** Gender: M Patient ID: 20220711230258006735	Specimen: GS006771A Collected: 11/15/2023 Received: 11/22/2023 / 16:47 EDT Reported: 10/30/2024 / 13:49 EDT	Client #: 97554902 MOTA, CHRISTINA

Tay-Sachs Disease

Result

NO CLINICALLY RELEVANT VARIANTS DETECTED

Clinical Relevance

No clinically relevant variants associated with Tay-Sachs disease were detected in the HEXA gene. As a screening test, this assay is not intended to rule out all variants in the gene(s) tested.

Tested Genes Include:
HEXA

Reviewer

Vivekananda Datta, MD, PhD

Methods and Limitations

This screening test should not be used for diagnosis without confirmation by other medically established means. When a clinically relevant variant(s) is identified, confirmatory testing is strongly recommended prior to making any clinical decisions. A negative result from this analysis does not rule out the possibility that the tested individual carries a rare unexamined variant, variant in an undetectable region, or a type of variant not intended to be detected by the test. For complete methods and limitations, please refer to the end of this report. If applicable, condition specific additional information can be found below.

Condition Specific Additional Information

This assay excludes detection of the presence or absence of the pseudodeficiency alleles in HEXA.

Healthcare Provider Resources

For general information about this test, visit www.questdiagnostics.com/genetic-health-screening

Patient Information	Specimen Information	Client Information
WARDEN, CHARLES **DOB: 04/05/1985 AGE: 38** Gender: M Patient ID: 20220711230258006735	Specimen: GS006771A Collected: 11/15/2023 Received: 11/22/2023 / 16:47 EDT Reported: 10/30/2024 / 13:49 EDT	Client #: 97554902 MOTA, CHRISTINA

APC-Associated Hereditary Cancer

Result

NO CLINICALLY RELEVANT VARIANTS DETECTED

Clinical Relevance

No clinically relevant variants associated with APC-associated hereditary cancer were detected in the APC gene. As a screening test, this assay is not intended to rule out all variants in the gene(s) tested.

Tested Genes Include:
APC

Reviewer

Vivekananda Datta, MD, PhD

Methods and Limitations

This screening test should not be used for diagnosis without confirmation by other medically established means. When a clinically relevant variant(s) is identified, confirmatory testing is strongly recommended prior to making any clinical decisions. A negative result from this analysis does not rule out the possibility that the tested individual carries a rare unexamined variant, variant in an undetectable region, or a type of variant not intended to be detected by the test. For complete methods and limitations, please refer to the end of this report. If applicable, condition specific additional information can be found below.

Condition Specific Additional Information

The intended use of this screening test is to identify pathogenic and likely pathogenic variants in the listed genes associated with an increased risk of developing APC-associated hereditary cancer, which may include, but is not limited to familial adenomatous polyposis (FAP), attenuated familial adenomatous polyposis (AFAP).

Healthcare Provider Resources

For general information about this test, visit www.questdiagnostics.com/genetic-health-screening

Patient Information	Specimen Information	Client Information
WARDEN, CHARLES **DOB: 04/05/1985 AGE: 38** Gender: M Patient ID: 20220711230258006735	Specimen: GS006771A Collected: 11/15/2023 Received: 11/22/2023 / 16:47 EDT Reported: 10/30/2024 / 13:49 EDT	Client #: 97554902 MOTA, CHRISTINA

ATM-Associated Hereditary Cancer

Result

NO CLINICALLY RELEVANT VARIANTS DETECTED

Clinical Relevance

No clinically relevant variants associated with ATM-associated hereditary cancer were detected in the ATM gene. As a screening test, this assay is not intended to rule out all variants in the gene(s) tested.

Tested Genes Include:
ATM

Reviewer

Vivekananda Datta, MD, PhD

Methods and Limitations

This screening test should not be used for diagnosis without confirmation by other medically established means. When a clinically relevant variant(s) is identified, confirmatory testing is strongly recommended prior to making any clinical decisions. A negative result from this analysis does not rule out the possibility that the tested individual carries a rare unexamined variant, variant in an undetectable region, or a type of variant not intended to be detected by the test. For complete methods and limitations, please refer to the end of this report. If applicable, condition specific additional information can be found below.

Healthcare Provider Resources

For general information about this test, visit www.questdiagnostics.com/genetic-health-screening

Patient Information	Specimen Information	Client Information
WARDEN, CHARLES **DOB: 04/05/1985 AGE: 38** Gender: M Patient ID: 20220711230258006735	Specimen: GS006771A Collected: 11/15/2023 Received: 11/22/2023 / 16:47 EDT Reported: 10/30/2024 / 13:49 EDT	Client #: 97554902 MOTA, CHRISTINA

CHEK2-Associated Hereditary Cancer

Result

NO CLINICALLY RELEVANT VARIANTS DETECTED

Clinical Relevance

No clinically relevant variants associated with CHEK2-associated hereditary cancer were detected in the CHEK2 gene. As a screening test, this assay is not intended to rule out all variants in the gene(s) tested.

Tested Genes Include:
CHEK2

Reviewer

Vivekananda Datta, MD, PhD

Methods and Limitations

This screening test should not be used for diagnosis without confirmation by other medically established means. When a clinically relevant variant(s) is identified, confirmatory testing is strongly recommended prior to making any clinical decisions. A negative result from this analysis does not rule out the possibility that the tested individual carries a rare unexamined variant, variant in an undetectable region, or a type of variant not intended to be detected by the test. For complete methods and limitations, please refer to the end of this report. If applicable, condition specific additional information can be found below.

Healthcare Provider Resources

For general information about this test, visit www.questdiagnostics.com/genetic-health-screening

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WARDEN, CHARLES **DOB: 04/05/1985 AGE: 38** Gender: M Patient ID: 20220711230258006735	Specimen: GS006771A Collected: 11/15/2023 Received: 11/22/2023 / 16:47 EDT Reported: 10/30/2024 / 13:49 EDT	Client #: 97554902 MOTA, CHRISTINA

Hereditary Breast and Ovarian Cancer

Result

NO CLINICALLY RELEVANT VARIANTS DETECTED

Clinical Relevance

No clinically relevant variants associated with hereditary breast and ovarian cancer syndrome were detected in the BRCA1 or BRCA2 genes. As a screening test, this assay is not intended to rule out all variants in the gene(s) tested.

Tested Genes Include:
BRCA1, BRCA2

Reviewer

Vivekananda Datta, MD, PhD

Methods and Limitations

This screening test should not be used for diagnosis without confirmation by other medically established means. When a clinically relevant variant(s) is identified, confirmatory testing is strongly recommended prior to making any clinical decisions. A negative result from this analysis does not rule out the possibility that the tested individual carries a rare unexamined variant, variant in an undetectable region, or a type of variant not intended to be detected by the test. For complete methods and limitations, please refer to the end of this report. If applicable, condition specific additional information can be found below.

Healthcare Provider Resources

For general information about this test, visit www.questdiagnostics.com/genetic-health-screening

Patient Information	Specimen Information	Client Information
WARDEN, CHARLES **DOB: 04/05/1985 AGE: 38** Gender: M Patient ID: 20220711230258006735	Specimen: GS006771A Collected: 11/15/2023 Received: 11/22/2023 / 16:47 EDT Reported: 10/30/2024 / 13:49 EDT	Client #: 97554902 MOTA, CHRISTINA

Juvenile Polyposis Syndrome (BMPR1A-associated)

Result

NO CLINICALLY RELEVANT VARIANTS DETECTED

Clinical Relevance

No clinically relevant variants associated with juvenile polyposis syndrome (BMPR1A-associated) were detected in the BMPR1A gene. As a screening test, this assay is not intended to rule out all variants in the gene(s) tested.

Tested Genes Include:
BMPR1A

Reviewer

Vivekananda Datta, MD, PhD

Methods and Limitations

This screening test should not be used for diagnosis without confirmation by other medically established means. When a clinically relevant variant(s) is identified, confirmatory testing is strongly recommended prior to making any clinical decisions. A negative result from this analysis does not rule out the possibility that the tested individual carries a rare unexamined variant, variant in an undetectable region, or a type of variant not intended to be detected by the test. For complete methods and limitations, please refer to the end of this report. If applicable, condition specific additional information can be found below.

Healthcare Provider Resources

For general information about this test, visit www.questdiagnostics.com/genetic-health-screening

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WARDEN, CHARLES **DOB: 04/05/1985 AGE: 38** Gender: M Patient ID: 20220711230258006735	Specimen: GS006771A Collected: 11/15/2023 Received: 11/22/2023 / 16:47 EDT Reported: 10/30/2024 / 13:49 EDT	Client #: 97554902 MOTA, CHRISTINA

Juvenile Polyposis Syndrome / Hereditary Hemorrhagic Telangiectasia (SMAD4-associated)

Result

NO CLINICALLY RELEVANT VARIANTS DETECTED

Clinical Relevance

No clinically relevant variants associated with juvenile polyposis syndrome/hereditary hemorrhagic telangiectasia (SMAD4-associated) were detected in the SMAD4 gene. As a screening test, this assay is not intended to rule out all variants in the gene(s) tested.

Tested Genes Include:
SMAD4

Reviewer

Vivekananda Datta, MD, PhD

Methods and Limitations

This screening test should not be used for diagnosis without confirmation by other medically established means. When a clinically relevant variant(s) is identified, confirmatory testing is strongly recommended prior to making any clinical decisions. A negative result from this analysis does not rule out the possibility that the tested individual carries a rare unexamined variant, variant in an undetectable region, or a type of variant not intended to be detected by the test. For complete methods and limitations, please refer to the end of this report. If applicable, condition specific additional information can be found below.

Healthcare Provider Resources

For general information about this test, visit www.questdiagnostics.com/genetic-health-screening

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Lynch Syndrome

Result

NO CLINICALLY RELEVANT VARIANTS DETECTED

Clinical Relevance

No clinically relevant variants associated with Lynch syndrome were detected in the MLH1, MSH2, MSH6 or PMS2 genes. As a screening test, this assay is not intended to rule out all variants in the gene(s) tested.

Tested Genes Include:
MLH1, MSH2, MSH6, PMS2

Reviewer

Vivekananda Datta, MD, PhD

Methods and Limitations

This screening test should not be used for diagnosis without confirmation by other medically established means. When a clinically relevant variant(s) is identified, confirmatory testing is strongly recommended prior to making any clinical decisions. A negative result from this analysis does not rule out the possibility that the tested individual carries a rare unexamined variant, variant in an undetectable region, or a type of variant not intended to be detected by the test. For complete methods and limitations, please refer to the end of this report. If applicable, condition specific additional information can be found below.

Condition Specific Additional Information

This assay excludes analysis of exons 11-15 in the PMS2 gene.

Healthcare Provider Resources

For general information about this test, visit www.questdiagnostics.com/genetic-health-screening

Patient Information	Specimen Information	Client Information
WARDEN, CHARLES **DOB: 04/05/1985 AGE: 38** Gender: M Patient ID: 20220711230258006735	Specimen: GS006771A Collected: 11/15/2023 Received: 11/22/2023 / 16:47 EDT Reported: 10/30/2024 / 13:49 EDT	Client #: 97554902 MOTA, CHRISTINA

MUTYH-Associated Polyposis

Result

NO CLINICALLY RELEVANT VARIANTS DETECTED

Clinical Relevance

No clinically relevant variants associated with MUTYH-associated polyposis were detected in the MUTYH gene. As a screening test, this assay is not intended to rule out all variants in the gene(s) tested.

Tested Genes Include:
MUTYH

Reviewer

Vivekananda Datta, MD, PhD

Methods and Limitations

This screening test should not be used for diagnosis without confirmation by other medically established means. When a clinically relevant variant(s) is identified, confirmatory testing is strongly recommended prior to making any clinical decisions. A negative result from this analysis does not rule out the possibility that the tested individual carries a rare unexamined variant, variant in an undetectable region, or a type of variant not intended to be detected by the test. For complete methods and limitations, please refer to the end of this report. If applicable, condition specific additional information can be found below.

Healthcare Provider Resources

For general information about this test, visit www.questdiagnostics.com/genetic-health-screening

Patient Information	Specimen Information	Client Information
WARDEN, CHARLES **DOB: 04/05/1985 AGE: 38** Gender: M Patient ID: 20220711230258006735	Specimen: GS006771A Collected: 11/15/2023 Received: 11/22/2023 / 16:47 EDT Reported: 10/30/2024 / 13:49 EDT	Client #: 97554902 MOTA, CHRISTINA

PALB2-Associated Hereditary Cancer

Result

NO CLINICALLY RELEVANT VARIANTS DETECTED

Clinical Relevance

No clinically relevant variants associated with PALB2-associated hereditary cancer were detected in the PALB2 gene. As a screening test, this assay is not intended to rule out all variants in the gene(s) tested.

Tested Genes Include:
PALB2

Reviewer

Vivekananda Datta, MD, PhD

Methods and Limitations

This screening test should not be used for diagnosis without confirmation by other medically established means. When a clinically relevant variant(s) is identified, confirmatory testing is strongly recommended prior to making any clinical decisions. A negative result from this analysis does not rule out the possibility that the tested individual carries a rare unexamined variant, variant in an undetectable region, or a type of variant not intended to be detected by the test. For complete methods and limitations, please refer to the end of this report. If applicable, condition specific additional information can be found below.

Healthcare Provider Resources

For general information about this test, visit www.questdiagnostics.com/genetic-health-screening

Patient Information	Specimen Information	Client Information
WARDEN, CHARLES **DOB: 04/05/1985 AGE: 38** Gender: M Patient ID: 20220711230258006735	Specimen: GS006771A Collected: 11/15/2023 Received: 11/22/2023 / 16:47 EDT Reported: 10/30/2024 / 13:49 EDT	Client #: 97554902 MOTA, CHRISTINA

Peutz-Jeghers Syndrome

Result

NO CLINICALLY RELEVANT VARIANTS DETECTED

Clinical Relevance

No clinically relevant variants associated with Peutz-Jeghers syndrome were detected in the STK11 gene. As a screening test, this assay is not intended to rule out all variants in the gene(s) tested.

Tested Genes Include:
STK11

Reviewer

Vivekananda Datta, MD, PhD

Methods and Limitations

This screening test should not be used for diagnosis without confirmation by other medically established means. When a clinically relevant variant(s) is identified, confirmatory testing is strongly recommended prior to making any clinical decisions. A negative result from this analysis does not rule out the possibility that the tested individual carries a rare unexamined variant, variant in an undetectable region, or a type of variant not intended to be detected by the test. For complete methods and limitations, please refer to the end of this report. If applicable, condition specific additional information can be found below.

Healthcare Provider Resources

For general information about this test, visit www.questdiagnostics.com/genetic-health-screening

Patient Information	Specimen Information	Client Information
WARDEN, CHARLES **DOB: 04/05/1985 AGE: 38** Gender: M Patient ID: 20220711230258006735	Specimen: GS006771A Collected: 11/15/2023 Received: 11/22/2023 / 16:47 EDT Reported: 10/30/2024 / 13:49 EDT	Client #: 97554902 MOTA, CHRISTINA

POLD1/POLE-Associated Hereditary Cancer

Result

NO CLINICALLY RELEVANT VARIANTS DETECTED

Clinical Relevance

No clinically relevant variants associated with POLD1/POLE-associated hereditary cancer were detected in the POLD1 or POLE genes. As a screening test, this assay is not intended to rule out all variants in the gene(s) tested.

Tested Genes Include:
POLD1, POLE

Reviewer

Vivekananda Datta, MD, PhD

Methods and Limitations

This screening test should not be used for diagnosis without confirmation by other medically established means. When a clinically relevant variant(s) is identified, confirmatory testing is strongly recommended prior to making any clinical decisions. A negative result from this analysis does not rule out the possibility that the tested individual carries a rare unexamined variant, variant in an undetectable region, or a type of variant not intended to be detected by the test. For complete methods and limitations, please refer to the end of this report. If applicable, condition specific additional information can be found below.

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Classical Ehlers-Danlos Syndrome

Result

NO CLINICALLY RELEVANT VARIANTS DETECTED

Clinical Relevance

No clinically relevant variants associated with classical Ehlers-Danlos syndrome were detected in the COL5A1 or COL5A2 genes. As a screening test, this assay is not intended to rule out all variants in the gene(s) tested.

Tested Genes Include:
COL5A1, COL5A2

Reviewer

Vivekananda Datta, MD, PhD

Methods and Limitations

This screening test should not be used for diagnosis without confirmation by other medically established means. When a clinically relevant variant(s) is identified, confirmatory testing is strongly recommended prior to making any clinical decisions. A negative result from this analysis does not rule out the possibility that the tested individual carries a rare unexamined variant, variant in an undetectable region, or a type of variant not intended to be detected by the test. For complete methods and limitations, please refer to the end of this report. If applicable, condition specific additional information can be found below.

Healthcare Provider Resources

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WARDEN, CHARLES **DOB: 04/05/1985 AGE: 38** Gender: M Patient ID: 20220711230258006735	Specimen: GS006771A Collected: 11/15/2023 Received: 11/22/2023 / 16:47 EDT Reported: 10/30/2024 / 13:49 EDT	Client #: 97554902 MOTA, CHRISTINA

Familial Thoracic Aortic Aneurysm and Dissection

Result

NO CLINICALLY RELEVANT VARIANTS DETECTED

Clinical Relevance

No clinically relevant variants associated with familial thoracic aortic aneurysm and dissection were detected in the ACTA2 gene. As a screening test, this assay is not intended to rule out all variants in the gene(s) tested.

Tested Genes Include:
ACTA2

Reviewer

Vivekananda Datta, MD, PhD

Methods and Limitations

This screening test should not be used for diagnosis without confirmation by other medically established means. When a clinically relevant variant(s) is identified, confirmatory testing is strongly recommended prior to making any clinical decisions. A negative result from this analysis does not rule out the possibility that the tested individual carries a rare unexamined variant, variant in an undetectable region, or a type of variant not intended to be detected by the test. For complete methods and limitations, please refer to the end of this report. If applicable, condition specific additional information can be found below.

Condition Specific Additional Information

This assay excludes analysis of exon 9 in the ACTA2 gene.

Healthcare Provider Resources

For general information about this test, visit www.questdiagnostics.com/genetic-health-screening

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WARDEN, CHARLES **DOB: 04/05/1985 AGE: 38** Gender: M Patient ID: 20220711230258006735	Specimen: GS006771A Collected: 11/15/2023 Received: 11/22/2023 / 16:47 EDT Reported: 10/30/2024 / 13:49 EDT	Client #: 97554902 MOTA, CHRISTINA

Loeys-Dietz Syndrome

Result

NO CLINICALLY RELEVANT VARIANTS DETECTED

Clinical Relevance

No clinically relevant variants associated with Loeys-Dietz syndrome were detected in the TGFB2, TGFB3, TGFBR1 or TGFBR2 genes. As a screening test, this assay is not intended to rule out all variants in the gene(s) tested.

Tested Genes Include:
TGFB2, TGFB3, TGFBR1, TGFBR2

Reviewer

Vivekananda Datta, MD, PhD

Methods and Limitations

This screening test should not be used for diagnosis without confirmation by other medically established means. When a clinically relevant variant(s) is identified, confirmatory testing is strongly recommended prior to making any clinical decisions. A negative result from this analysis does not rule out the possibility that the tested individual carries a rare unexamined variant, variant in an undetectable region, or a type of variant not intended to be detected by the test. For complete methods and limitations, please refer to the end of this report. If applicable, condition specific additional information can be found below.

Healthcare Provider Resources

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WARDEN, CHARLES **DOB: 04/05/1985 AGE: 38** Gender: M Patient ID: 20220711230258006735	Specimen: GS006771A Collected: 11/15/2023 Received: 11/22/2023 / 16:47 EDT Reported: 10/30/2024 / 13:49 EDT	Client #: 97554902 MOTA, CHRISTINA

Marfan Syndrome

Result

NO CLINICALLY RELEVANT VARIANTS DETECTED

Clinical Relevance

No clinically relevant variants associated with Marfan syndrome were detected in the FBN1 gene. As a screening test, this assay is not intended to rule out all variants in the gene(s) tested.

Tested Genes Include:
FBN1

Reviewer

Vivekananda Datta, MD, PhD

Methods and Limitations

This screening test should not be used for diagnosis without confirmation by other medically established means. When a clinically relevant variant(s) is identified, confirmatory testing is strongly recommended prior to making any clinical decisions. A negative result from this analysis does not rule out the possibility that the tested individual carries a rare unexamined variant, variant in an undetectable region, or a type of variant not intended to be detected by the test. For complete methods and limitations, please refer to the end of this report. If applicable, condition specific additional information can be found below.

Healthcare Provider Resources

For general information about this test, visit www.questdiagnostics.com/genetic-health-screening

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WARDEN, CHARLES **DOB: 04/05/1985 AGE: 38** Gender: M Patient ID: 20220711230258006735	Specimen: GS006771A Collected: 11/15/2023 Received: 11/22/2023 / 16:47 EDT Reported: 10/30/2024 / 13:49 EDT	Client #: 97554902 MOTA, CHRISTINA

Vascular Ehlers-Danlos Syndrome

Result

NO CLINICALLY RELEVANT VARIANTS DETECTED

Clinical Relevance

No clinically relevant variants associated with vascular Ehlers-Danlos syndrome were detected in the COL3A1 gene. As a screening test, this assay is not intended to rule out all variants in the gene(s) tested.

Tested Genes Include:
COL3A1

Reviewer

Vivekananda Datta, MD, PhD

Methods and Limitations

This screening test should not be used for diagnosis without confirmation by other medically established means. When a clinically relevant variant(s) is identified, confirmatory testing is strongly recommended prior to making any clinical decisions. A negative result from this analysis does not rule out the possibility that the tested individual carries a rare unexamined variant, variant in an undetectable region, or a type of variant not intended to be detected by the test. For complete methods and limitations, please refer to the end of this report. If applicable, condition specific additional information can be found below.

Healthcare Provider Resources

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Methods and Limitations

Methods: This test is intended for detection of single nucleotide variants (SNVs) and small insertions and deletions (indels) in genes covered by the assay. Genomic DNA is sheared and libraries are constructed via hybrid capture. Libraries are sequenced on an Illumina NovaSeq6000 using paired-end 150 base pair reads. Reference genome alignment (hg38), variant calling, variant annotation, and variant quality filtering are performed using a bioinformatics pipeline that has been internally developed and validated. Final interpretation of results are performed by a board-certified geneticist.

Limitations: The intended use of this screening test is to identify pathogenic and likely pathogenic variants in the listed genes associated with the condition(s) above. For certain conditions, the analysis is restricted to determining whether specific variants noted above are present or absent. Variants of uncertain significance, likely benign and benign variants will not be reported. Compound heterozygous variants are assumed to exist with one variant present on each chromosome. Confirmatory testing is recommended in situations where variant(s) are found, given the screening nature of this test. This screening test is not designed to detect all variants that can cause the condition above and additional variants may significantly contribute to the condition examined. For example, not all variant types, such as copy number variants (CNVs) and rearrangements, are detected by this technology. As such, a negative result from the analysis cannot rule out the possibility that the tested individual carries a variant in the tested genes. Moreover, this test does not analyze additional genes associated with the condition above other than those specified. Therefore, results should be interpreted in the context of clinical findings, relevant history, and other laboratory data. Genetic counseling and discussion of these results with a physician is recommended. In some situations, additional genetic testing may be appropriate.

The test is not intended to detect copy number variants (CNVs) or other aberrations including changes to gene expression, epigenetic modifications, gene fusion, chromosome conformational changes, genomic rearrangements, skewed X-inactivation, and other unknown abnormalities. It does not detect SNVs, indels, or variants in regions of the gene not analyzed. Such regions include gene promoters, 5' and 3' untranslated regions, introns, and certain regions having consistently low coverage; thus, the test is not intended to detect intronic or intergenic variants. This test may not detect variants in patients exhibiting mosaicism. Mosaic variants may be reported, or missed, dependent upon the underlying mosaic fraction. Regions with exceptionally high or low GC content, repetitive regions, or regions having a high degree of similarity with other regions in the genome, are more susceptible to low coverage and/or reduced accuracy. Regions that did not meet quality criteria were not evaluated for the presence or absence of variants. In addition, the effect of rare or novel variants on mRNA splicing, protein synthesis, and/or protein function may remain unclear. Although rare, false positive or false negative results may occur. A negative result from the analysis cannot rule out the possibility that the tested individual carries a rare unexamined variant or variant in an undetectable region.

The classification and interpretation of the variant(s) identified reflect the current state of Quest Diagnostics' understanding at the time of this report. Variant classification and interpretation are subject to professional judgment, and may change for a variety of reasons, including but not limited to, updates in classification guidelines and availability of additional scientific and clinical information. This test result should be used in conjunction with confirmatory testing, if indicated, and a health care provider's clinical evaluation. Confirmatory testing as well as inquiry regarding potential changes to the classification of the variant(s) is strongly recommended prior to making any clinical decision. For questions regarding variant classification updates, providers may contact Quest Genomics Client Services at 1.866.GENE.INFO (1.866.436.3463).

Additional Information

Providers may contact Quest Genomics Client Services at 1.866.GENE.INFO (1.866.436.3463) for assistance with result interpretation, questions about variant classification, or to discuss additional testing.

This test was developed and its analytical performance characteristics have been determined by Quest Diagnostics. It has not been cleared or approved by the FDA. This assay has been validated pursuant to the CLIA regulations and is used for clinical purposes. This test should not be used for diagnosis without confirmation by other medically established means.

Performing Site

Athena Diagnostics, 200 Forest Street, 2nd Floor, Marlborough, MA, 01752-3023, Laboratory Director: Vivekananda Datta, MD, PhD, CLIA# 22D0069726