Patient ID / MRN	Patient Name		Birth Date	Gender	Age
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Genetic Health Screen (NGS)

This test evaluates 167 genes associated with various hereditary cancers, cardiovascular diseases, and other diseases for which effective medical interventions and preventive measures are available. See the Appendix below for the details of the gene-disease association.

Variants of uncertain significance are not included in this report; however, if additional evidence becomes available in the future that changes a previously classified variant of uncertain significance to a likely pathogenic or pathogenic variant, an updated report will be issued.

RESULT

Pathogenic Variant is Detected
MYH7:Chr14(GRCh37):g.23898214; NM_000257.4(MYH7):c.1357C>T; p.Arg453Cys; exon 34/40; heterozygous; coverage 75X

A heterozygous pathogenic variant c.1357C>T; p.Arg453Cys in the MYH7 gene was detected in this individual. This result strongly supports the molecular diagnosis of hypertrophic cardiomyopathy (HCM) in this individual. Therefore, the use of clinical practice guidelines (such as American College of Cardiology (ACC)/American Heart Association (AHA) guidelines, PMID: 33215938) is recommended for the diagnosis, treatment, and management of HCM in this individual, and assessment of genetic risk and cardiac surveillance of at-risk relatives.

INTERPRETATION

MYH7:Chr14(GRCh37):g.23898214; NM_000257.4(MYH7):c.1357C>T; p.Arg453Cys

Normal gene/protein function: Myosin muscle is a hexameric protein containing 2 heavy chain subunits, 2 alkali light chain subunits, and 2 regulatory light chain subunits. The MYH7 gene encodes the beta (or slow) heavy chain subunit of cardiac myosin. It is expressed predominantly in the normal human ventricle. It is also expressed in skeletal muscle tissues rich in slow-twitch type I muscle fibers. Changes in the relative abundance of this protein and the alpha (or fast) heavy subunit of cardiac myosin correlate with the contractile velocity of cardiac muscle. Its expression is also altered during thyroid hormone depletion and hemodynamic overloading.

Mutation effect: Pathogenic variants in the MYH7 gene are associated with hypertrophic cardiomyopathy (autosomal dominant/digenic dominant, OMIM: 192600), dilated cardiomyopathy (autosomal dominant, OMIM: 613426), Laing Distal Myopathy (autosomal dominant, OMIM: 160500), myosin storage congenital myopathy (autosomal dominant/recessive, OMIM 608358/255160) and Left ventricular noncompaction (autosomal dominant, OMIM: 613426).

The variant c.1357C>T; p.Arg453Cys in the MYH7 gene alters a highly conserved amino acid and a meta-predictor (REVEL) predicts it as damaging to the protein function. This variant is located on the Myosin head, the motor domain of the MYH7 protein (InterPro). This variant is present in the general population from gnomAD with an allele frequency of 0.0003% (6/1,614,098 alleles, no homozygotes). This variant has been recurrently reported in individuals with Hypertrophic Cardiomyopathy or Restrictive Cardiomyopathy including evidence of cosegregation with disease and do novo occurrences (PMID: 8655135, 11133230, 12084606, 12881443, 12951062, 12975413, 15358028, 15856146). Functional studies have shown

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that this variant affects MYH7 function (PMID: 15001446, 17351073, 23798412, 24344137). Several clinical laboratories in ClinVar have classified this variant as Pathogenic (ClinVar ID: 14089). Based on the available evidence, this variant has been classified as Pathogenic.

METHODOLOGY

The total genomic DNA was extracted from the biological sample using the CE-IVD-marked Bead-based EZ1 DSP DNA Blood Kit (Qiagen, Hilden, Germany). After assessment of DNA quality and quantity using the Denovix DS-11 Spectrophotometer/Fluorometer system, the DNA was randomly fragmented, and ligated sequencing libraries were prepared using Twist Library Preparation EF 2.0 kit (Twist Biosciences, CA, USA). Regions of interest (coding exons and flanking intronic regions) were targeted by the hybridization-based target capture method using the Twist Exome 2.0 kit (Twist Biosciences). Captured DNA was sequenced to an average targeted depth of ≥100X on the Illumina NextSeq 2000 using 2x150 bp paired-end reads (Illumina, San Diego, CA, USA). Primary data analysis converting images into base calls and associated quality scores and secondary analysis aligning the sequencing reads against the reference human genome (GRCh37-hg19) and variant calling was carried out by Illumina's proprietary software (Dragen 3.10.12). Copy number variation (CNV) calling was performed using ClinCNV (1.18.3), Dragen CNV (3.10.12), and VS-CNV (2.6.2). Structural variation (SV) calling was performed using Dragen SV (3.10.12) and Delly (1.2.6). Repeat Expansion calling was performed by Expansion Hunter in Dragen 3.10.12. Variant annotation and filtering were performed on VarSeq 2.6.2 (Golden Helix, MT, USA). Variant interpretation was performed based on the published ACMG/AMP guidelines (Richard et al, 2015, PMID: 25741868) along with ClinGen gene/disease specifications. To avoid pseudogene interference, any reportable variant in the PMS2 gene is confirmed by long-range PCR.

PANEL GENE LIST (n=167)

ABCD1, ACTA2, ACTC1, ACTN2, ACVRL1, AIP, APC (including promoter region), APOB, ATM, ATP7B, AXIN2, BAG3, BAP1, BARD1, BMPR1A, BMPR2, BRCA1, BRCA2, BRIP1, BTD, CACNA1C, CACNA1S, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV1, CAV3, CDC73, CDH1, CDK4, CDKN1B, CDKN2A, CHEK2, COL3A1, COL5A1, COL5A2, CRYAB, CSRP3, DES, DICER1, DMD, DSC2, DSG2, DSP, EGFR, EMD, ENG, EPCAM (copy number variation only), F2, F5, F9, FBN1, FH, FHL1, FLCN, FLNC, G6PD, GAA, GCH1, GDF2, GLA, GPD1L, GREM1 (copy number variation only), HAMP, HCN4, HFE, HJV, HMBS, HNF1A, HNF1B, HOXB13, JUP, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, KIT, LAMP2, LDLR, LDLRAP1, LMNA, LZTR1, MAX, MEFV, MEN1, MET, MITF, MLH1, MSH2, MSH3, MSH6, MUTYH, MYBPC3, MYH11, MYH7, MYL2, MYL3, MYLK, NF1, NF2, NKX2-5, NTHL1, OTC, PALB2, PCSK9, PDGFRA, PKP2, PLN, PMS2, POLD1, POLE, POT1, PRKAG2, PRKAR1A, PRKG1, PROC, PROS1, PTCH1, PTEN, RAD51C, RAD51D, RB1, RBM20, RET, RPE65, RYR1, RYR2, SCN5A, SDHA, SDHAF2, SDHB, SDHC, SDHD, SERPINA1, SERPINC1, SGCD, SLC40A1, SMAD3, SMAD4, SMAD9, SMARCA4, SMARCB1, STK11, TCAP, TFR2, TGFB2, TGFB3, TGFBR1, TGFBR2, TMEM127, TMEM43, TNNC1, TNNI3, TNNT2, TP53, TPM1, TRDN, TSC1, TSC2, TTN, TTR, VCL, VHL, WT1

COVERAGE METRICS

Coverage of target region ≥10X in 99.99%
Coverage of target region ≥20X in 99.91%
Coverage of target region ≥30X in 98.96%

A minimum coverage of ≥10X for each target/gene was used as the cut-off as the passing criteria. Any target region in a gene not meeting this cut-off is noted under the LIMITATIONS section.

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PERFORMANCE CHARACTERISTICS

Single base substitution: accuracy >99%; reproducibility 100% (intra- and inter-assay). Small insertion/deletion events (up to 50 bp): accuracy >99%; reproducibility 100% (intra- and inter-assay). Larger single gene insertions/deletions with size ≥50bp that primarily encompass single to multi-exons will also be reported.

LIMITATIONS

Next-generation sequencing may not detect all types of genomic variants. This test does not detect complex inversions, gene conversions, balanced translocations, repeat expansion disorders, and noncoding variants deeper than ±20 base pairs from the exon-intron boundary unless otherwise indicated. Additionally, this test may not reliably detect the following: low-level mosaicism, variants in the stretch of mononucleotide repeats, indels larger than 50bp, and single exon deletions or duplications. False-negative results may also occur in the setting of bone marrow transplantation, recent blood transfusion, or suboptimal DNA quality. In addition, the chance of a false positive or false negative result due to rare laboratory errors incurred during any phase of testing cannot be completely excluded. The depth of coverage may be variable for some target regions, but assay performance below the minimum acceptable criteria will be noted. In addition, some exons and intronic regions within ±20 base pairs are not analyzed due to inherent low quality due to homology/pseudogene and repetitive regions. Given these limitations, negative results do not rule out the diagnosis of a genetic disorder.

Exon/exonic regions not analyzed: exon 15 in PMS2 and exon 172-197 in TTN. These exons are excluded due to homology/pseudogene interference.
Intron ± 20 bp not analyzed (but ±2 bp included): None

DISCLAIMER

This is a screening test only; therefore, it does not include all the genes related to the genetic diseases tested. This test was developed, and its performance characteristics were determined by the Molecular Diagnostics & Genomics Laboratory of NRL. The test is currently not accredited by the Emirates International Accreditation Centre (EIAC) but will be added to the scope of accreditation for the next assessment cycle.

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APPENDIX

1. Hereditary cancer genes (n=65)

GENE	ASSOCIATED DISEASE	GENE	ASSOCIATED DISEASE
AIP	Pituitary cancer	MUTYH	Colorectal cancer
APC	Colorectal cancer, Gastric cancer	NF1	Nervous system/brain cancer, Breast cancer, Gastric cancer
ATM	Breast cancer, Pancreatic cancer, Prostate cancer	NF2	Nervous system/brain cancer
AXIN2	Colorectal cancer	NTHL1	Colorectal cancer, Breast cancer
BAP1	BAP1 tumor predisposition syndrome	PALB2	Breast cancer, Pancreatic cancer, Ovarian cancer, Prostate cancer
BARD1	Breast cancer	PDGFRA	Gastrointestinal cancer
BMPR1A	Colorectal cancer, Gastric cancer	PMS2	Colorectal cancer, Endometrial cancer, Ovarian cancer, Prostate cancer, Pancreatic cancer, Gastric cancer
BRCA1	Breast cancer, Ovarian cancer, Prostate cancer, Pancreatic cancer	POLD1	Colorectal cancer
BRCA2	Breast cancer, Ovarian cancer, Prostate cancer, Pancreatic cancer	POLE	Colorectal cancer
BRIP1	Ovarian cancer	POT1	POT1 tumor predisposition syndrome
CDC73	Endocrine cancer, Renal cancer	PRKAR1A	Endocrine cancer, Nervous system/brain cancer
CDH1	Breast cancer, Gastric cancer	PTCH1	Nervous system/brain cancer

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CDK4	Melanoma, Pancreatic cancer	PTEN	Breast cancer, Endometrial cancer, Colorectal cancer, Thyroid cancer
CDKN1B	Multiple endocrine neoplasia type 4	RAD51C	Ovarian cancer, Breast cancer
CDKN2A	Melanoma, Pancreatic cancer	RAD51D	Ovarian cancer, Breast cancer
CHEK2	Breast cancer, Colorectal cancer, Prostate cancer	RB1	Retinoblastoma
DICER1	Lung cancer, Thyroid cancer, Ovarian cancer	RET	Multiple endocrine neoplasia 2
EGFR	Lung cancer	SDHA	Endocrine cancer, Gastrointestinal cancer
EPCAM	Colorectal cancer	SDHAF2	Endocrine cancer
FH	Renal cancer, Urinary tract cancer, Endocrine cancer	SDHB	Endocrine cancer, Gastrointestinal cancer, Renal cancer
FLCN	Kidney cancer	SDHC	Endocrine cancer, Gastrointestinal cancer
GREM1	Colorectal cancer	SDHD	Endocrine cancer, Gastrointestinal cancer
HOXB13	Prostate cancer	SMAD4	Colorectal cancer, Gastric cancer, Pancreatic cancer
KIT	Gastrointestinal cancer	SMARCA4	Rhabdoid tumor predisposition syndrome, Ovarian cancer
LZTR1	Neuronal cancer	SMARCB1	Nervous system/brain cancer, Renal cancer
MAX	Endocrine cancer	STK11	Breast cancer, ovarian cancer, pancreatic cancer, endometrial cancer, gastric cancer, colorectal cancer
MEN1	Multiple endocrine neoplasia 1	TMEM127	Endocrine cancer
MET	Renal cancer	TP53	Breast cancer, Brain cancer, Endocrine cancer, Renal cancer, Ovarian cancer, Colorectal cancer, Gastric cancer, Prostate cancer, Pancreatic cancer
MITF	Melanoma	TSC1	Nervous system/brain cancer, Pancreatic cancer, Renal/urinary tract cancer
MLH1	Colorectal cancer, Endometrial cancer, Ovarian cancer, Prostate cancer, Pancreatic cancer, Gastric cancer	TSC2	Nervous system/brain cancer, Pancreatic cancer, Renal/urinary tract cancer
MSH2	Colorectal cancer, Endometrial cancer, Ovarian cancer, Prostate cancer, Pancreatic cancer, Gastric cancer	VHL	Endocrine cancer, Nervous system/brain cancer, Pancreatic cancer, renal/urinary tract cancer
MSH3	Colorectal cancer	WT1	Renal/urinary tract cancer
MSH6	Colorectal cancer, Endometrial cancer, Ovarian cancer, Prostate cancer, Pancreatic cancer, Gastric cancer		

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2. Hereditary cardiovascular disease genes (n=83)

GENE	ASSOCIATED DISEASE	GENE	ASSOCIATED DISEASE
ACTA2	Aortopathy	KCNQ1	Long QT syndrome, Atrial fibrillation, Jervell and Lange-Nielsen syndrome, Short QT syndrome
ACTC1	Cardiomyopathy, Congenital heart disease	LAMP2	Danon disease
ACTN2	Arrhythmia, Cardiomyopathy	LDLR	Familial hypercholesterolemia
ACVRL1	Hereditary hemorrhagic telangiectasia, Pulmonary arterial hypertension	LDLRAP1	Familial hypercholesterolemia
APOB	Familial Hypercholesterolemia, Familial Hypobetalipoproteinemia	LMNA	Cardiomyopathy, Emery-Dreifuss muscular dystrophy, Charcot-Marie-Tooth disease, Hutchinson-Gilford progeria syndrome
BAG3	Cardiomyopathy, Myofibrillar myopathy	MYBPC3	Cardiomyopathy, Left ventricular non-compaction
BMPR2	Pulmonary arterial hypertension	MYH11	Aortopathy
CACNA1C	Arrhythmia, Cardiomyopathy, Congenital heart disease	MYH7	Cardiomyopathy, Congenital myopathy, Laing distal myopathy, Left ventricular non-compaction
CACNB2	Arrhythmia	MYL2	Cardiomyopathy, Myofibrillar myopathy
CALM1	Arrhythmia	MYL3	Cardiomyopathy
CALM2	Arrhythmia	MYLK	Aortopathy
CALM3	Arrhythmia	NKX2-5	Arrhythmia, Congenital heart disease
CASQ2	Arrhythmia	PCSK9	Familial hypercholesterolemia
CAV1	Pulmonary arterial hypertension	PKP2	Arrhythmia
CAV3	Arrhythmia, Cardiomyopathy, Distal myopathy, Rippling muscle disease	PLN	Cardiomyopathy
COL3A1	Ehlers-Danlos syndrome	PRKAG2	Cardiomyopathy, Arrhythmia
COL5A1	Ehlers-Danlos syndrome	PRKG1	Aortopathy
COL5A2	Ehlers-Danlos syndrome	PROC	Thrombophilia
CRYAB	Cardiomyopathy, Myofibrillar myopathy	PROS1	Thrombophilia
CSRP3	Cardiomyopathy	RBM20	Arrhythmia, Cardiomyopathy
DES	Cardiomyopathy, Myofibrillar myopathy	RYR2	Arrhythmia, Cardiomyopathy
DMD	Cardiomyopathy, Muscular dystrophy	SCN5A	Arrhythmia, Cardiomyopathy
DSC2	Arrhythmia	SERPINC1	Thrombophilia
DSG2	Arrhythmia, Cardiomyopathy	SGCD	Cardiomyopathy, Limb-girdle muscular dystrophy

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DSP	Arrhythmias, Cardiomyopathy, Epidermolysis bullosa, Keratosis palmoplantaris striata	SMAD3	Aortopathy
EMD	Muscular dystrophy	SMAD4	Hereditary hemorrhagic telangiectasia
ENG	Hereditary hemorrhagic telangiectasia	SMAD9	Pulmonary arterial hypertension
F2	Thrombophilia	TCAP	Cardiomyopathy, Muscular dystrophy
F5	Thrombophilia	TGFB2	Aortopathy
F9	Thrombophilia	TGFB3	Aortopathy, Arrhythmia, Cardiomyopathy
FBN1	Marfan syndrome, Heritable thoracic aortic disease, Geleophysic dysplasia, Weill-Marchesani syndrome	TGFBR1	Aortopathy
FHL1	Myopathy	TGFBR2	Aortopathy
FLNC	Cardiomyopathy, Myopathy, Arrhythmia	TMEM43	Arrhythmia, Cardiomyopathy
GDF2	Hereditary hemorrhagic telangiectasia	TNNC1	Cardiomyopathy
GLA	Fabry disease	TNNI3	Arrhythmia, Cardiomyopathy
GPD1L	Arrhythmia	TNNT2	Arrhythmia, Cardiomyopathy
HCN4	Arrhythmia	TPM1	Cardiomyopathy
JUP	Arrhythmogenic right ventricular dysplasia, Naxos disease	TRDN	Arrhythmia
KCNE1	Long QT syndrome, Jervell and Lange-Nielsen syndrome	TTN	Cardiomyopathy, Myofibrillar myopathy, Muscular dystrophy
KCNE2	Long QT syndrome, Atrial fibrillation	TTR	Systemic amyloidosis (includes cardiomyopathy)
KCNH2	Long QT syndrome	VCL	Cardiomyopathy
KCNJ2	Atrial fibrillation, Andersen syndrome, Short QT syndrome		

3. Other genes (n=20)

GENE	ASSOCIATED DISEASE	GENE	ASSOCIATED DISEASE
ABCD1	X-linked adrenoleukodystrophy	HMBS	Acute intermittent porphyria
ATP7B	Wilson disease	HNF1A	Maturity-onset diabetes of the young (MODY)
BTD	Biotinidase deficiency	HNF1B	Maturity-onset diabetes of the young (MODY), Renal cysts and diabetes syndrome
CACNA1S	Hypokalemic periodic paralysis, Malignant hyperthermia susceptibility	MEFV	Familial Mediterranean fever
G6PD	Glucose-6-phosphate dehydrogenase deficiency	OTC	Ornithine transcarbamylase deficiency
GAA	Glycogen storage disease	RPE65	Retinal dystrophy
GCH1	Dystonia, Hyperphenylalaninemia	RYR1	Malignant hyperthermia Susceptibility, Myopathy

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HAMP	Hereditary Hemochromatosis	SERPINA1	Alpha-1-antitrypsin deficiency
HFE	Hereditary hemochromatosis	SLC40A1	Hereditary hemochromatosis
HJV	Hereditary hemochromatosis	TFR2	Hereditary hemochromatosis

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