Patient name: example report
HN: 123456789
Date of birth: 02/01/1985
Sex: Male

Sample type: EDTA Blood
Specimen id: 12345678-1
Date of collection: 01/03/2022
Date of receive: 01/03/2022

Date of result: 28/04/2023
Physician order: Dr. Examplereport Test

RESULT: Negative

TEST INFORMATION

Cardio screening includes 98 genes as shown in the section "condition associated gene" below.

TEST RESULTS

No genetic variant with clinical significance is found.

INTERPRETATION SUMMARY

There were no known, clinically significant genetic changes detected that confer a genetic predisposition to, or carrier status for, certain types of heart conditions analyzed in this panel. Please refer to the complete list of genes and conditions below. Please also note that other risks based on non-genetic factors or other genetic causes not evaluated with this test may still be of clinical significance.

RECOMMENDATIONS

The interpretation of these results should be done in the context of a patient's medical record and family history. Please note that interpretation and classification of the variants reported here may change over time. Please see a genetic counselor for services regarding the implications of these results in the context of understanding the implications of incidental findings, family planning and the informing of family members of potentially shared genetic outcomes.

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METHODOLOGY

Genomic DNA is extracted from an individual at Bumrungrad Hospital. DNA sample is sent to the Macrogen, Korea to process Whole Genome Sequencing (WGS). Library preparation, clustering and sequencing are processed on the Illumina platform. Data in a mean depth of 30X were generated. Reads from the sequence output were aligned to the human reference genome (GRCh37) and processed for variant calling (SNP/Indel) using the Illumina pipeline (Isaac.v4). Manta is performed to identify structural variants and large indels while copy number variant is identified by Control-FREEC. The tertiary analysis is performed at Bumrungrad Hospital. The variants were annotated and filtered using the Golden Helix VarSeq analysis workflow implementing the ACMG guidelines for the interpretation of sequence variants. This includes a comparison against the gnomAD population catalog of variants in 123,136 exomes, the 1000 Genomes Project Consortium's publication of 2,500 genomes, the NCBI ClinVar database of clinical assertions on variant's pathogenicity and multiple lines of computational evidence on conservation and functional impact.

Coverage Statistics for cardio screen panel

Target region / Coverage	WGS Target region	Cardio panel target region
Mean depth (X)	**30.9X**	**29.90X**
Mean depth ≥ 10X	**98.8%**	**99.71%**

VARIANT ASSESSMENT PROCESS

The following databases and in-silico algorithms are used to annotate and evaluate the impact of the variant in the context of human disease: 1000 genomes, gnomAD, ClinVar, OMIM, dbSNP, NCIB RefSeq Genes, ExAC Gene Constraints, VS-SIFT, VS-PolyPhen2, PhyloP, GERP++, GeneSplicer, MaxEntScan, NNSplice, PWM Splice Predictor. Analysis was reported using the to HGVS nomenclature (www.hgvs.org/mutnomen) as implemented by the VarSeq transcript annotation algorithm. The reported transcript matches that used most frequently by the clinical labs submitting to ClinVar.

LIMITATIONS

It should be noted that the test result is limited to a set of genes indicated in the panel and might not cover all possible variants related to the particular condition. For some target regions, the depth covered for analysis may be variable, However, any targeted gene that fails to meet the acceptance criteria (Mean depth □ 10X) will be noted. Due to these limitations, ruling out the diagnosis of a genetic disorder should not be made based on negative results. An evaluation by alternative methods should be considered if a specific clinical disorder is suspected. This report only includes variants that meet a level of evidence threshold for cause or contribute to disease/condition. Reported variants are not confirmed by Sanger sequencing. Certain classes of genomic variants are also not covered using the NGS testing technology, including repeat expansions, large deletion or large duplication (□50 kb), translocations and gene fusions or other complex structural rearrangements.

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DISCLAIMER

The result interpretation is based on the most current scientific and analytical standards. However, more evidence for disease association of genes and causal pathogenic variants are discovered every year, and it is recommended that genetic variants are re-interpreted with updated software and annotations periodically. There is also a possibility of an error in the result due to contaminants in the sample, rare technical errors, a rare genetic variant that could interfere with the analysis. This test should be used in compliance with the other diagnostic test. Note that this test cannot exclude the possibility of variants in genes not analyzed or assayed with incomplete coverage. Even though this test is not designed to distinguish between somatic and germline variants, if variant of somatic is detected, supplementary testing may be compulsory to clarify the significance of results. Genetic counseling is recommended to help understand the test result and explain the implications of this result for the patients and other family members. This test has not been cleared or approved by the U.S. Food and Drug Administration (FDA). The FDA has determined that such clearance or approval is not necessary.

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CONDITION ASSOCIATED GENE

The table shows the list of 98 genes related to cardiovascular conditions analyzed in this test. Gene-Phenotype relationship information is retrieved from http://www.omim.org.

Gene	Transcript	Gene MIM number	Condition associated gene
ACTA2	NM_001613.4	102620	Aortic aneurysm, familial thoracic 6, Moyamoya disease 5, Multisystemic smooth muscle dysfunction syndrome
ACTC1	NM_005159.5	102540	Atrial septal defect, Dilated cardiomyopathy, Hypertrophic cardiomyopathy, Left ventricular noncompaction
ACTN2	NM_001103.4	102573	Dilated cardiomyopathy, Hypertrophic cardiomyopathy, Distal Myopathy
ACVRL1	NM_000020.3	601284	Hereditary hemorrhagic telangiectasia
ANKRD1	NM_014391.3	609599	Dilated cardiomyopathy
APOB	NM_000384.3	107730	Hypercholesterolemia, Hypobetalipoproteinemia
BAG3	NM_004281.4	603883	Dilated cardiomyopathy, Myofibrillar myopathy
BMPR2	NM_001204.7	600799	Pulmonary hypertension, Pulmonary veno-occlusive disease
CACNA1C	NM_000719.7	114205	Brugada syndrome, Long QT syndrome, Timothy syndrome
CACNB2	NM_201590.3	600003	Brugada syndrome
CALM1	NM_006888.6	114180	Long QT syndrome, Catecholaminergic polymorphic ventricular tachycardia
CALM2	NM_001743.6	114182	Long QT syndrome
CALM3	NM_005184.4	114183	Catecholaminergic polymorphic ventricular tachycardia, Long QT syndrome
CASQ2	NM_001232.4	114251	Catecholaminergic polymorphic ventricular tachycardia
CAV1	NM_001753.5	601047	Primary pulmonary hypertension
CAV3	NM_033337.3	601253	Hypertrophic cardiomyopathy, Long QT syndrome, Distal Myopathy
COL3A1	NM_000090.4	120180	Vascular Ehlers-Danlos syndrome
CRYAB	NM_001885.3	123590	Dilated cardiomyopathy, Myofibrillar Myopathy
CSRP3	NM_003476.5	600824	Dilated cardiomyopathy, Hypertrophic cardiomyopathy
DES	NM_001927.4	125660	Dilated cardiomyopathy, Myofibrillar Myopathy
DMD	NM_004006.3	300377	Dilated cardiomyopathy, Muscular dystrophy
DSC2	NM_024422.6	125645	Arrhythmogenic right ventricular Cardiomyopathy
DSG2	NM_001943.5	125671	Arrhythmogenic right ventricular Cardiomyopathy, Dilated cardiomyopathy
DSP	NM_004415.4	125647	Arrhythmogenic right ventricular Cardiomyopathy, Dilated cardiomyopathy
DTNA	NM_001390.4	601239	Left ventricular noncompaction
EMD	NM_000117.3	300384	Muscular dystrophy
ENG	NM_000118.3	131195	Hereditary hemorrhagic telangiectasia
EYA4	NM_004100.5	603550	Dilated cardiomyopathy
F2	NM_000506.5	176930	Prothrombin deficiency, Thrombophilia
F5	NM_000130.5	612309	Factor V deficiency, Thrombophilia
F9	NM_000133.4	300746	Hemophilia, Thrombophilia
FBN1	NM_000138.5	134797	Geleophysic dysplasia, Marfan syndrome, MASS syndrome,Weill-Marchesani syndrome
FHL1	NM_001449.5	300163	Uruguay faciocardiomusculoskeletal syndrome, Muscular dystrophy
FKTN	NM_001079802.2	607440	Dilated cardiomyopathy, Muscular dystrophy

Patient name: Anuree Sitachitt **HN**: 100002566

Gene	Transcript	Gene MIM number	Condition associated gene
FLNC	NM_001458.5	102565	Hypertrophic cardiomyopathy, Familial restrictive cardiomyopathy, Distal myopathy, Myofibrillar myopathy
GATAD1	NM_021167.5	614518	Dilated cardiomyopathy
GDF2	NM_016204.4	605120	Hereditary hemorrhagic telangiectasia
GLA	NM_000169.3	300644	Fabry disease
GPD1L	NM_015141.4	611778	Brugada syndrome
HCN4	NM_005477.3	605206	Brugada syndrome, Sick sinus syndrome
JPH2	NM_020433.5	605267	Dilated cardiomyopathy, Hypertrophic cardiomyopathy
JUP	NM_002230.4	173325	Arrhythmogenic right ventricular Cardiomyopathy, Naxos disease
KCNE1	NM_000219.6	176261	Jervell and Lange-Nielsen syndrome, Long QT syndrome
KCNE2	NM_172201.2	603796	Familial atrial fibrillation, Long QT syndrome
KCNH2	NM_000238.4	152427	Long QT syndrome, Short QT syndrome
KCNJ2	NM_000891.3	600681	Andersen syndrome, Familial atrial fibrillation, Short QT syndrome
KCNQ1	NM_000218.3	607542	Familial atrial fibrillation, Jervell and Lange-Nielsen syndrome, Long QT syndrome
LAMA4	NM_002290.5	600133	Dilated cardiomyopathy
LAMP2	NM_002294.3	309060	Danon disease
LDB3	NM_007078.3	605906	Dilated cardiomyopathy, Hypertrophic cardiomyopathy, Left ventricular noncompaction, Myofibrillar myopathy
LDLR	NM_000527.5	606945	Familial Hypercholesterolemia
LDLRAP1	NM_015627.3	605747	Familial Hypercholesterolemia
LMNA	NM_170707.4	150330	Dilated cardiomyopathy, Muscular dystrophy, Heart-hand syndrome, Hutchinson-Gilford progeria syndrome, Familial partial lipodystrophy, Malouf syndrome
MAP2K1	NM_002755.4	176872	Cardiofaciocutaneous syndrome
MAP2K2	NM_030662.4	601263	Cardiofaciocutaneous syndrome
MYBPC3	NM_000256.3	600958	Dilated cardiomyopathy, Hypertrophic cardiomyopathy, Left ventricular noncompaction
MYH11	NM_002474.3	160745	Familial thoracic aortic aneurysm and dissection
MYH6	NM_002471.4	160710	Atrial septal defect, Dilated cardiomyopathy, Hypertrophic cardiomyopathy, Sick sinus syndrome
MYH7	NM_000257.4	160760	Dilated cardiomyopathy, Hypertrophic cardiomyopathy, Distal myopathy, Left ventricular noncompaction, Myopathy
MYL2	NM_000432.4	160781	Cardiomyopathy, Myofibrillar myopathy
MYL3	NM_000258.3	160790	Hypertrophic cardiomyopathy
MYLK	NM_053025.4	600922	Familial thoracic aortic aneurysm and dissection
MYLK2	NM_033118.4	606566	Hypertrophic cardiomyopathy
MYOZ2	NM_016599.5	605602	Hypertrophic cardiomyopathy
MYPN	NM_032578.4	608517	Dilated cardiomyopathy, Familial restrictive cardiomyopathy, Hypertrophic cardio, Myopathy
NEXN	NM_144573.4	613121	Dilated cardiomyopathy, Hypertrophic cardiomyopathy
NKX2-5	NM_004387.4	600584	Atrial septal defect, Conotruncal heart malformations, Hypoplastic left heart syndrome, Tetralogy of Fallot, Ventricular septal defect
PCSK9	NM_174936.4	607786	Familial hypercholesterolemia
PKP2	NM_004572.4	602861	Arrhythmogenic right ventricular Cardiomyopathy
PLN	NM_002667.5	172405	Dilated cardiomyopathy, Hypertrophic cardiomyopathy
PRKAG2	NM_016203.4	602743	Hypertrophic cardiomyopathy, Lethal congenital glycogen storage disease of the heart, Wolff-Parkinson-White syndrome

Patient name: Anuree Sitachitt **HN**: 100002566

Gene	Transcript	Gene MIM number	Condition associated gene
PRKG1	NM_006258.4	176894	Familial thoracic aortic aneurysm and dissection
PROC	NM_000312.4	612283	Thrombophilia
PROS1	NM_000313.4	176880	Thrombophilia
RAF1	NM_002880.4	164760	Dilated cardiomyopathy, LEOPARD syndrome, Noonan syndrome
RBM20	NM_001134363.3	613171	Dilated cardiomyopathy
RYR2	NM_001035.3	180902	Arrhythmogenic right ventricular Cardiomyopathy, Ventricular arrhythmias, Catecholaminergic polymorphic ventricular tachycardia
SCN5A	NM_198056.3	600163	Familial atrial fibrillation, Brugada syndrome, Dilated cardiomyopathy, Progressive familial heart block, Long QT syndrome, Sick sinus syndrome, Familial ventricular fibrillation
SERPINC1	NM_000488.4	107300	Thrombophilia
SGCD	NM_000337.6	601411	Dilated cardiomyopathy, Muscular dystrophy
SMAD3	NM_005902.4	603109	Loeys-Dietz syndrome
SMAD4	NM_005359.6	600993	Hereditary hemorrhagic telangiectasia
TA2	NM_000116.5	300394	Barth syndrome
TCAP	NM_003673.4	604488	Hypertrophic cardiomyopathy, Muscular dystrophy
TGFB2	NM_003238.6	190220	Loeys-Dietz syndrome
TGFB3	NM_003239.5	190230	Arrhythmogenic right centricular cardiomyopathy, Loeys-Dietz syndrome
TGFBR1	NM_004612.4	190181	Loeys-Dietz syndrome
TGFBR2	NM_003242.6	190182	Loeys-Dietz syndrome
TMEM43	NM_024334.3	612048	Arrhythmogenic right ventricular cardiomyopathy, Muscular dystrophy
TMPO	NM_003276.2	188380	Dilated cardiomyopathy
TNNC1	NM_003280.3	191040	Dilated cardiomyopathy, Hypertrophic cardiomyopathy
TNNI3	NM_000363.5	191044	Dilated cardiomyopathy, Hypertrophic cardiomyopathy, Familial restrictive cardiomyopathy
TNNT2	NM_001001430.3	191045	Dilated cardiomyopathy, Familial restrictive cardiomyopathy, Hypertrophic cardiomyopathy, Left ventricular noncompaction
TPM1	NM_001018005.2	191010	Dilated cardiomyopathy, Hypertrophic cardiomyopathy, Left ventricular noncompaction
TRDN	NM_006073.4	603283	Cardiac arrhythmia syndrome
TTN	NM_001267550.2	188840	Dilated cardiomyopathy, Hypertrophic cardiomyopathy, Muscular dystrophy, Myofibrillar myopathy
TTR	NM_000371.4	176300	Hereditary transthyretin(TTR)-related amyloidosis
VCL	NM_014000.3	193065	Dilated cardiomyopathy, Hypertrophic cardiomyopathy