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

TEST INFORMATION

This test is a screening tool to determine the risks of developing Cardiovascular disease and Alzheimer's disease by analyzing 2 SNP markers (rs429358 and rs7412)

VARIANTS RELEVANT TO INDICATION FOR TESTING

Gene	Chromosome position	rsID	Variant	Interpretation
APOE	19:45411941	rs429358	Homozygous T/C	**APOE E2/E4**
APOE	19:45412079	rs7412	Homozygous T/C	

Interpretation for Cardiovascular disease:

This genotype is associated with normal lipid metabolism, and is not associated with an increased risk of cardiovascular disease.

Interpretation for Alzheimer's disease:

This genotype is associated with increased risk of developing Alzheimer's disease compared to those with the APOE E3/E3 genotype.

References

1. Eichner JE, Dunn ST, Perveen G, Thompson DM, Stewart KE, Stroehla BC. Apolipoprotein E polymorphism and cardiovascular disease: a HuGE review. Am J Epidemiol. 2002 Mar 15;155(6):487-95.
2. Schaefer EJ, Lamon-Fava S, Johnson S, Ordovas JM, Schaefer MM, Castelli WP, Wilson PW. Effects of gender and menopausal status on the association of apolipoprotein E phenotype with plasma lipoprotein levels. Results from the Framingham Offspring Study. Arterioscler Thromb. 1994 Jul;14(7):1105-13.
3. Song Y, Stampfer MJ, Liu S. Meta-analysis: apolipoprotein E genotypes and risk for coronary heart disease. Ann Intern Med. 2004 Jul 20;141(2):137-47.
4. Goldman JS, Hahn SE, Catania JW, LaRusse-Eckert S, Butson MB, Rumbaugh M, Strecker MN, Roberts JS, Burke W, Mayeux R, Bird T; American College of Medical Genetics and the National Society of Genetic Counselors. Genetic counseling and testing for Alzheimer disease: joint practice guidelines of the American College of Medical Genetics and the National Society of Genetic Counselors. Genet Med. 2011 Jun;13(6):597-605.
5. Van Cauwenberghe C, Van Broeckhoven C, Sleegers K. The genetic landscape of Alzheimer disease: clinical implications and perspectives. Genet Med. 2016 May;18(5):421-30.
6. Li Z, Shue F, Zhao N, Shinohara M, Bu G. APOE2: protective mechanism and therapeutic implications for Alzheimer's disease. Mol Neurodegener. 2020 Nov 4;15(1):63.

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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.

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 interpretation of this test is based on a limited number of SNP and might not cover all possible variants related to the particular condition. Lifestyle and other factors that might affect the condition are not accounted on the analysis.

DISCLAIMER

The results of this test are not intended, and should not be used to diagnose or treat any disease or medical condition, but could provide useful information about how to manage any disease or conditions you may have holistically. There is also a possibility of an error in the result due to contaminants in the sample, rare technical errors, and a rare genetic variant that could interfere with the analysis. 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.