SG GENETIC DISEASES DIAGNOSTIC CENTER
MEDICAL ANALYSIS REPORT
Laboratory license number: GHDM-SM/34.39/01

Patient Name	:		**Sampling Location**	:	
Gender	:		**Test Request Date**	:	
Date Of Birth	:		**Sample Collection Date**	:	
ID Number	:		**Sample Acceptance Date**	:	
Protocol Number	:		**Report Approval Date**	:	
Sample Number	:		**Report Release Date/Report Number**	:	/ 00
Sample Type	:				

Refering Center / Physician :

Reason For Referral :

Test name: Whole Exome Sequencing (WES)
Analysis method: Next Generation Sequencing (NGS)
Platform: Illumina NovaSeq 6000
Kit used: Twist Human Core Exome Kit, Seqscape, Twist Bioscience, Sophia, Mastermind
Bioinformatic analysis: Sophia DDM v4

Scope of the analysis: In this study, DNA obtained from patient's peripheral blood samples were used and the coding regions (exons) and exon/intron junctions and genes listed in HPO (The Human Phenotype Ontology) phenotypes included in the analysis were examined and 81 genes[1] belonging to the incidental findings in line with the recommendations of ACMG (American College of Medical Genetics) were filtered in detail in line with the shared clinical information, and pathogenic/probably pathogenic/suspicious variants of unknown clinical significance that could be compatible with family history and clinic were listed. Genome-wide ClinVar pathogenic findings were also filtered and included in the analysis in line with the shared clinical information.

[1]The gene panel content associated with ACMG secondary findings (81 Genes) is indicated in the report.

DATA ANALYSIS RESULTS

POSITIVE,

HOMOZYGOUS LIKELY PATHOGENIC2 MISSENSE VARIANT WAS DETECTED in GCDH GENE.

[2]The variant class is determined according to the ACMG criteria.

Variant Table

Gene (Transcript)	Variant	Zygosity	dbSNP	Variant Class	Inheritance3
GCDH (NM_000159.3)	-	-	-	-	-

[3] Inheritance; AR: Autosomal recessive, AD: Autosomal dominant

INTERPRETATION:

As a result of the whole exome sequencing (WES) studied on DNA material obtained from patient's peripheral blood homozygous missense c.1202T>A (p.Ile401Asn) variant was detected in GCDH (NM_000159.3) gene which was classified as likely pathogenic. ClinVar database doesn't classify this variant. dbSNP database has no ID for this variant. Minor Allele Frequency (MAF) of this variant is not shared. In OMIM database pathogenic variants in…. gene are associated with (OMIM: #) phenotypes (OMIM: #).

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SECONDARY FINDINGS:

We did not detect any pathogenic/ likely pathogenic variant in the 81 genes for which incidental findings are reported based on the ACMG (American College of Medical Genetics and Genomics) Secondary Findings Manual (David T Miller et al., 2023) with the consent of the patient.

ACMG SECONDARY FINDINGS GENE PANEL (81 GENES):

ACTA2, ACTC1, ACVRL1, APC, APOB, ATP7B, BAG3, BMPR1A, BRCA1, BRCA2, BTD, CACNA1S, CALM1, CALM2, CALM3, CASQ2, COL3A1, DES, DSC2, DSG2, DSP, ENG, FBN1, FLNC, GAA, GLA, HFE, HNF1A, KCNH2, KCNQ1, LDLR, LMNA, MAX, MEN1, MLH1, MSH2, MSH6, MUTYH, MYBPC3, MYH11, MYH7, MYL2, MYL3, NF2, OTC, PALB2, PCSK9, PKP2, PMS2, PRKAG2, PTEN, RB1, RBM20, RET, RPE65, RYR1, RYR2, SCN5A, SDHAF2, SDHB, SDHC, SDHD, SMAD3, SMAD4, STK11, TGFBR1, TGFBR2, TMEM127, TMEM43, TNNC1, TNNI3, TNNT2, TP53, TPM1, TRDN, TSC1, TSC2, TTN, TTR, VHL, WT1

RECOMMENDATIONS:

This section includes recommendations and follow up criteria according to the findings.

QUALITY CONTROL RESULTS:

Target region's average coverage:	X	**Target region's minimal 25X coverage:**	%
Target region's minimal 50X coverage:	%	**Target region's minimal 100X coverage:**	%

TECHNICAL SPECIFICATIONS and LIMITATIONS:

Information is provided about the specification and limitations of the kit and technique used. This section also includes information about what is included or excluded from the report within the scope of the study and analysis.

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REFERENCES:

1. Miller, David T et al. “ACMG SF v3.2 list for reporting of secondary findings in clinical exome and genome sequencing: A policy statement of the American College of Medical Genetics and Genomics (ACMG).” Genetics in medicine : official journal of the American College of Medical Genetics, 100866. 15 Jun. 2023, doi:10.1016/j.gim.2023.100866
2. Richards, Sue, et al. "Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology." Genetics in medicine 17.5 (2015): 405.

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