Clinical Whole Exome Sequencing Report-Solo

NoorDx test WES001

Patient Name:	Referring Physician
DOB:	Physician's Address:
Gender:	Contract No:
MRN / ID:	Specimen Type:
Client Code:	NoorDx Code:
Clinical indications:	

Negative: No clinically relevant variants identified

Recommendations:

- Clinical correlation and genetic counseling are highly recommended.
- Reanalysis of the sequence dataset is recommended every 12 months or if there are phenotypic changes.

ACMG Secondary Findings

Not opted for in the consent form.

Copy Number Variants (CNVs) Analysis:

No phenotype relevant CNVs were detected.

Quality metrics

Coverage: 112x	Reads covering variant: N/A	Confirmed by: N/A

The classification of the variations is done based on the guidelines of the American College of Medical Genetics as described below1:

	Description
Variant classification Pathogenic	A disease-causing variant in a gene which can explain the patient's symptoms has been detected. This usually means that a suspected disorder for which testing had been requested has been confirmed.
Likely pathogenic	A variant which is very likely to contribute to the development of disease however, the scientific evidence is currently insufficient to prove this conclusively. Additional evidence is expected to confirm this assertion of pathogenicity.

Variant of Uncertain Significance (VOUS)	A variant has been detected, but it is difficult to classify it as either pathogenic (disease causing) or benign (non-disease causing) based on current available scientific evidence. Further testing of the patient or family members as recommended by your clinician may be needed. It is probable that their significance can be assessed only with time, subject to availability of scientific evidence.

Test Method:

This test was developed and validated by NoorDx Laboratory. This test has been developed for clinical purposes. All the test results are reviewed, interpreted, and reported by licensed genetic practitioners. In line with ACMG standards and guidelines for the interpretation of sequence variants[2], secondary findings are reported or reviewed during this analysis upon the request of the patient and/health provider.

The sequencing was performed on genomic DNA using targeted exome enrichment. In-house clinically validated tools and utilities including bioinformatics pipeline, base calling, primary filtering of low-quality reads and probable artifacts, and annotation of variants was applied. Variants filtration and classification are performed in-house. All the phenotypically relevant VOUS, likely pathogenic and pathogenic variants in ClinVar, HGMD, as well as all variants with minor allele frequency (MAF) of less than 1% in ExAc, ESP, gnomAD, SNP and in-house database in addition to any variant lies between exons and intron boundaries +/-20 has been considered. CNV predictions were performed using bioinformatic tools that includes Germline CNV Caller from GATK and Control-Freec (v11.6). All relevant inheritance patterns based on provided family history and clinical information are used to evaluate identified variants. Genomic variants are for hg38 assembly. Only variants related to the phenotype are reported. All reported variants are validated by Sanger sequencing.

Test Limitation:

Variants lies outside the coding regions of covered genes or failing to meet quality control scores might not be detected. Furthermore, some genetics changes may not be identified since next-generation sequencing technologies cannot precisely read through repeat expansion regions and thus cannot yield data i.e., for repeat expansion in cases with Fragile X syndrome or other repeat expansion disorders in other regions and or copy number variations (CNVs). Copy number variations (CNVs), defined as single exon or larger deletions or duplications (Del/Dups), were analyzed from the sequence analysis data using a validated proprietary bioinformatics pipeline. However, CNV analysis from WES data remains a screening test that requires orthogonal confirmatory assays like chromosomal microarray, karyotyping, FISH or MLPA analyses. Test results are interpreted in the context of clinical findings and family history indicated by the physician in the request form.

Only variations in known or candidate (if opted for) genes that are potentially related to the proband's medical conditions are reported. Rare polymorphisms may lead to false negative or positive results. False positive variants may arise through sequencing and bioinformatic selection/filtering artefacts (i.e., the variants do not exist within the patient exome but are artificially elicited during the sequencing/computational analysis) and will likely be excluded by the Sanger confirmation. False negative results may arise due to standard bioinformatic filtering conditions for variant selection. Clinical variant reporting and classification may change in light of newly acquired information (e.g., new clinical information, updated family history, and newly published relevant literature). If the obtained results do not match the clinical findings, additional testing should be considered.

Disclaimer:

This test does not detect the following: complex inversions, gene conversions, balanced translocations, repeat expansion disorders, non-coding and intronic variants deeper than ±20 base pairs from exon-intron boundary. Additionally, this test may not reliably detect the following: low level mosaicism, stretches of mononucleotide repeats, indels larger than 50bp, and variants within pseudogene regions/duplicated segments. A negative result does not rule out the diagnosis of a genetic disorder since some DNA abnormalities may be undetectable by the applied technology. Test results should always be interpreted in the context of clinical findings, family history, and other relevant data. Inaccurate, or incomplete information may lead to misinterpretation of the results.

Regulatory Disclosure:

This report has been produced by NoorDx Diagnostic and Discovery Laboratory, Kingdom of Saudi Arabia. The laboratory is accredited, by the Ministry of Health in Saudi Arabia, for molecular testing. The information contained within this report should ONLY be used in conjunction with other clinical results associated with the patient. The data should not be used as the sole diagnostic tool.

References:

[1]Richards, S. et al. Genet Med. 2015; 17(5):405-24

Signed:

Manar Samman, Ph.D.
Molecular Genetics Consultant

Naif Almontashiri, Ph.D, DABMGG, FACMG
Molecular Genetics Consultant and Chief Genetics Officer

Noor Diagnostics and Discovery
4017 Wadi Lane, Thuwal, KSA
0128083814 – 0128087982
info@noordx.sa | noordx.sa

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Patient Name:
Specimen No.
MRN:

Procedure	Specimen #	Collected	Received	Reported
WES-Solo		05/12/2022 10:38 AM	05/12/2022 04:00 PM	01/01/2023 02:30 PM

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