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GENOME DIAGNOSTICS REPORT

Mw. dr. S. Albert
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Alhier

Our reference: R24-04479 / 23-01148

Nijmegen, 29-03-2024

PERSONAL DETAILS

Name:	S-240209-00583
Date of Birth:	11-11-1800
Sex:	U
Date of request:	22-02-2024
Indication:	CNV analyse in WES data (WES)
Reason for referral:	

SAMPLE DETAILS

Material	Collection date	Date of receipt	DNA-number
DNA	unknown	22-02-2024	DNA24-03096

RESULTS AND MOLECULAR INTERPRETATION

Exome wide Copy Number Variant (CNV) analysis in the exome sequencing data did NOT reveal any (likely) causative CNVs. Normal female CNV profile.

CONCLUSION

A normal female CNV profile was observed.

REMARKS

CoNIFER: 67 segments; 24/67 with Z-score >|1,6|.

This test was performed using the provided DNA sample labelled S-240209-00583.

TEST DESCRIPTION

Exome sequencing was performed with Illumina NovaSeq 6000, after exome enrichment with the Twist Exome 2.0 plus Comprehensive Exome Spike-in Kit. Read alignment was done with BWA, and variant calling with CoNIFER. Subsequently, variant were annotated by the department of Human Genetics of the Radboudumc using an in-house developed pipeline.
Confirmation of reported CNV's was only performed when explicitly stated in the report.

DISCLAIMER

The exome sequencing that was performed, covers a large part of the exome (all exones of the genome). On this data, genome-wide CNV analysis was performed in which the CNV detection software needs a minimum of three aberrant targets (exons) to detect a CNV. Inherent to the structure of the human genome (such as the centromeric, heterochromatic and gene-poor regions) the resolution of this CNV detection varies. Balanced chromosome rearrangements cannot be detected with array (CGH) analysis. Low mosaic genomic imbalances may remain undetectable. The genomic positions that are stated in this report are based on Human Genome Build GRCh37/hg19 (February 2009).

With kind regards,

Ing. N.F.A. Leijsten
Senior Technician

Ms. Dr. N. de Leeuw
Clinical Laboratory Geneticist *

This report has been signed and authorised electronically (*).