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GENOME DIAGNOSTICS REPORT

Mw. dr. S. Albert
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Alhier

Our reference: R25-19883 / 23-01148

Nijmegen, 17-09-2025

PERSONAL DETAILS

Name:	S-250805-00761
Date of Birth:	11-11-1800
Sex:	U
Date of request:	05-08-2025
Indication:	CNV analysis in WES/WGS data
Reason for referral:	confirmation of the clinical diagnosis

SAMPLE DETAILS

Material	Collection date	Date of receipt	DNA-number
DNA	unknown	05-08-2025	DNA25-13094

RESULTS AND MOLECULAR INTERPRETATION

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

CONCLUSION

A normal female CNV profile was observed.

REMARKS

CoNIFER: 27 segments; 14/27 with Z-score >|1,6|.

This test was performed using the provided DNA sample labelled S-250805-00761.

TEST DESCRIPTION

Exome sequencing was performed with a Illumina NovaSeq 6000, after enrichment of the exome using the Twist Exome 2.0 plus Comprehensive Exome Spike-in Kit. "Read alignment" was performed with BWA, and "variant calling" with CoNIFER and/or ExomeDepth for copy number variants (CNVs). The variants were then annotated by the Genetics department of Radboudumc and MaastrichtUMC+ using an in-house developed pipeline. Reported CNVs are only confirmed with an independent test if explicitly stated in the results.

DISCLAIMER

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 (*).