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GENOME DIAGNOSTICS REPORT

Mw. dr. S. Albert
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Alhier

Our reference: R25-19886 / 23-01148

Nijmegen, 17-09-2025

PERSONAL DETAILS

Name:	S-250805-00758
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	DNA25-13091

RESULTS AND MOLECULAR INTERPRETATION

Abnormal female CNV profile.
With exome wide CNV analysis in the exome data an interstitial copy number gain of about 755 kb was detected in the distal long arm of chromosome 5. This concerns:

seq[GRCh38] 5q33.3(156757580_157509467)x3
NC_000005.10:g.(156647616_156757580)_(157509467_157513389)dup VUS

The gain in 5q33.3 encompasses 12 protein-coding genes and has not been detected / reported before with this size by us or others in any other patient. The proximal breakpoint of this gain is in the SGCD gene (OMIM *601411) and the distal breakpoint in ADAM19 (OMIM *603640). At this moment, this copy number gain is classified as variant of uncertain significance.

CONCLUSION

At this moment it is not clear whether the interstitial gain in 5q33.3 has clinical consequences.

REMARKS

CoNIFER: 166 segments; 85/166 with Z-score >|1,6|.

This test was performed using the provided DNA sample labeled S-250805-00758.

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 (*).