EXOME SEQUENCING REPORT

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Clinical geneticist

Our reference: X00-00000 / 00-00000
Your reference: 0000000000

Nijmegen, 00-00-0000

PERSONAL DETAILS

Name: X. XXXX
Date of Birth: 00-00-0000
Sex: M

Date of request: 00-00-0000
Indication: intellectual disability (gene panel analysis, NGS)
Reason for referral: confirmation of the clinical diagnosis

SAMPLE DETAILS

Material	Date of receipt	DNA-number
DNA	00-00-0000	DNA00-00000

RESULTS AND MOLECULAR INTERPRETATION

Using exome sequencing, NO causative pathogenic mutations were detected in the genes associated with intellectual disability (gene panel version DGD_14112014).

CONCLUSION

No genetic cause of the counselee's intellectual disability was identified using this test.
Since no clear pathogenic mutations were identified in the genes within the gene panel, the rest of the exome data will be analysed. If likely pathogenic mutations are identified with that strategy, a new report will be issued.

TEST DESCRIPTION

Exome enrichment (Agilent SureSelectXT Human all Exon 50 Mb), exome sequencing (Illumina HiSeq2000TM), read alignment (BWA) and variant calling (GATK) were done at BGI-Europe (Denmark). Variant annotation, selection, and prioritizing for pathogenicity was done by the department of Human genetics, Radboudumc, using an in-house developed strategy. This pipeline also filters the data for the genes defined in the gene panel, if applicable (information about the gene panel and its prioritization is available on request, and accessible from our website). Confirmation of reported variants by an independent technique (such as Sanger sequencing) has only been performed for low-quality variants (GATK quality scores). This has been validated to have a >99.9% reliability of the non-confirmed variants to be present and to be 'de novo' (if applicable).

DISCLAIMER

This exome sequencing test produces data that cover the majority of the exome. Nevertheless, some areas of the exome are poorly covered or absent from the data. Thus, mutations in those regions may remain unidentified. Furthermore, some types of mutations (such as repeat expansions, copy-number variants, mitochondrial-DNA mutations and areas beyond the exome, including introns and promoters) will not have been detected by this test. The gene panels are updated regularly, but may be incomplete due to the continuous identification of novel genes in human disease. Specific information about the coverage, gene panels, etc. is available on request. The aim of this test is to reveal the cause of the disorder in the counselee, not to reveal the putative carrier status of recessive disorders. Carrier status of a given disorder may be requested in particular situations, ie in case of consanguinity between the counselee and his/her partner, or if the partner of the counselee has a molecularly-confirmed disorder. Due to small likelihood (<1:1000) of a false-positive report (see test description), there is a small risk of a withdrawal of a previous conclusion. This should be considered during counseling of the proband, or family members (cascade screening). Especially in future prenatal testing, a prior request of confirmation of a mutation is indicated.

With kind regards,

Y. YYYY, PhD	Z.ZZZZ, PhD
Clinical Laboratory Geneticist	Clinical Laboratory Geneticist *

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