Whole Exome Sequencing on the Illumina Novaseq 6000 NGS Platofrm

| Clinical Indication: |
| --- |
| {d.report.clinicalSummary} |

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| --- |

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Variant Interpretation & Clinical correlation:

{d.report.FLNC}

{d.report.interpretation}

| Variant Evidence | Gene Impact |
| --- | --- |
| Chromosome: {d.chromosomeno}  Position: {d.position}  NC\_000017.11(GRCh38 CHr17):  {d.Nc}   | Allele | DP | % | | --- | --- | --- | | {d.Allele} | {d.DP} | {d.%} | |  |  |  |   Genotype: phred quality score:  {d.heterozygous} {d.phredscore} | RefSeq Gnese 110, NCBI   | Gene:  {d.gene} | Transcript:  {d.Transcript} | | --- | --- | | Effect:  {d.effect} | Protein:  {d.protein} | | Exon:  {d.exon} | Coding:  {d.coding} | |

Based on the evidence, this variant {d.FLNC} is classified as likely to be {d.pathogenic} variant

OMIM Phenotype:

{d.OMIMPhenotype}

| Case Specific Recommendations: |
| --- |
| {d.caseSpecificRecommendations} |

| Recommendations |
| --- |
| {d.recommendation} |

| Methodology: |
| --- |
| {d.test\_methodology} |

| Limitations/Disclaimer: |
| --- |
| {d.Disclaimer} |

Variant Classification as per ACMG guidelines:

|  |  |
| --- | --- |
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|  |  |

References:

{d.references}

Appendix 1: Sample Data and Statistics

| Title | Data |
| --- | --- |
| {d.title[i]} | {d.data[i]} |
| {d.title[i]} | {d.data[i]} |
| {d.title[i]} | {d.data[i]} |
| {d.title[i]} | {d.data[i]} |

Appendix 2: Coverage Summary

| Mean Depth | Percentage target base pairs covered |
| --- | --- |

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| --- | --- | --- | --- |
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Appendix 3: Coverage of Analyzed Genes (Percentage of coding region covered)

| Gene | Region Covered | Gene | Region Covered | Gene | Region Covered |
| --- | --- | --- | --- | --- | --- |
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