Whole Exome Sequencing on the Illumina Novaseq 6000 NGS Platofrm

| Clinical Indication: |
| --- |
| {d.dynamic.clinicalSummary.data} |

|  |
| --- |

\*

Variant Interpretation & Clinical correlation:

{d.dynamic.FLNC.data}

{d.dynamic.interpretation.data}

| Variant Evidence | Gene Impact |
| --- | --- |
| Chromosome: {d.dynamic.variantInfo.data.Location}  Position: {d.position}  {d.dynamic.variantInfo.data.Variation}   | Allele | DP | % | | --- | --- | --- | | {d.dynamic.variantInfo.data.rowData[0].Zygosity} | {d.dynamic.variantInfo.data.rowData[0].DP} | {d.dynamic.variantInfo.data.rowData[0].%} | |  |  |  |   Genotype:  {d.dynamic.variantInfo.data..Zygosity}  phred quality score: {d.dynamic.variantInfo.data.phredscore} | RefSeq Gnese 110, NCBI   | Gene:  {d.dynamic.variantInfo.data.Gene (Transcript)} | Transcript:  {d.dynamic.variantInfo.data.Gene (Transcript)} | | --- | --- | | Effect:  {d.dynamic.variantInfo.data.Classification} | Protein:  {d.dynamic.variantInfo.data.Variation} | | Exon:  {d.dynamic.variantInfo.data.Variation} | Coding:  {d.dynamic.variantInfo.data.Variation} | |

Based on the evidence, this variant {d.dynamic.variantInfo.data.rowData[0].Variation}

is classified as likely to be {d.dynamic.variantInfo.data.rowData[0].Classification}

variant

OMIM Phenotype

{d.dynamic.variantInfo.data.rowData[0].Disease (OMIM)}

| Case Specific Recommendations: |
| --- |
| {d.dynamic.recommendation.data} |

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

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

| Limitations/Disclaimer: |
| --- |
| {d.static.limitation.data}  {d.static.disclaimer.data} |

Variant Classification as per ACMG guidelines:

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

References:

{d.static.references.data}

Appendix 1: Sample Data and Statistics

{#d.dynamic.databaseInformation}

{description}

{#data.rowData}

{title} {data}

{/data.rowData}

{/d.dynamic.databaseInformation}

Appendix 2: Coverage Summary

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

| {d.coverageSummary.meanDepth} | {d.coverageSummary.percentageCovered} |  |  |
| --- | --- | --- | --- |
|  |  |  |  |

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

{#d.coverageOfAnalyzedGenes}

Gene: {Gene}

Region Covered: {Region Covered}

{/d.coverageOfAnalyzedGenes}

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