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# nucleome

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**Name Sex/Age**

**Date Received: Indication**

{d.dynamic.caseMetaData.proband.name}

{d.dynamic.caseMetaData.proband.sex}/{d.dynamic.caseMetaData.proband.age}

{d.dynamic.caseMetaData.proband.sampleReceived}

**Whole Exome Sequencing on the Illumina Novaseq 6000 NGS Platform**



-

{d.dynamic.clinicalSummary.data}

{d.dynamic.result.data}

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| Gene and Transcript | Location | Variant | Zygosity | OMIM Phenotype | Clinical Significance |
| {d.dynamic.variantInfo.data.rowData[0][2]} | {d.dynamic.variantInfo.data.rowData[0][2]} |  | {d.dynamic.variantInfo.data.rowData[0][4]} | {d.dynamic.variantInfo.data.rowData[0][6]} | {d.dynamic.clinicalSummary.data} |

*\*Genetic test results are reported based on the recommendations of American College of Medical Genetics*

**h** .

**FLCN:c.1150 1160delGTCCAGTCAGC:**

{d.dynamic.interpretation.data}

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Phred Quality Score:

44.00

1 ,n10,000 probability of FP

Genotype:

{d.dynamic.variantInfo.data.rowData[0][4]}

Depth:14

%

Allele DP

{d.dynamic.variantInfo.data.rowData[0][2]}

7

7

g.17217086\_17217096delCTGACTGGACG

Right aligned lbp to 17,217.086

Position:

{d.dynamic.variantInfo.data.rowData[0][1]}

{d.dynamic.variantInfo.data.rowData[0][2]}

{d.dynamic.variantInfo.data.rowData[0][2]}

{d.dynamic.variantInfo.data.rowData[0][2]}

{d.dynamic.variantInfo.data.rowData[0][2]}

Chromosome:

{d.dynamic.variantInfo.data.rowData[0][1]}

NC \_000017 11 IGRCh38 Chrl7):

I

**ll**◄

**GRCh37mm**

Variant Evidence for 2400146295.targeted

I

Variant Evidence



**07-Aug-2024**

Gene Impact

Gene:

*{d.dyanmic.variantInfo.data.rowData[0][1]}*

Effect:

{d.dynamic.variantInfo.data.rowData[0][6]}

Exon:

□□□□□□□□□■□□□□



{d.dynamic.variantInfo.data.rowData[0][1]}



ProtP,n:

Coding:

***Based on the evidence, this variant {d.dynamic.variantInfo.data.rowData[0][4]}***

***is classified as likely to be  {d.dynamic.variantInfo.data.rowData[0][6]}***

***variant***

**OMIM Phenotype:**

{d.dynamic.variantInfo.data.rowData[0][6]}



* {d.static.recommendation.data[0]}
* {d.static.recommendation.data[1]}
* {d.static.recommendation.data[2]}
* {d.static.recommendation.data[3]}

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Methodology

{d.static.test\_methodology.data}

Limitations/Disclaimer:

{d.static.limitation.data}

{d.static.disclaimer.data}



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Variant Classification as per ACMG

|  |  |
| --- | --- |
| Name | Description |
| {d.static.terms.data.rowData[0][0]} | {d.static.terms.data.rowData[0][1]} |
| {d.static.terms.data.rowData[1][0]} | {d.static.terms.data.rowData[1][1]} |

***References:***

1. ***{d.static.references.data[0]}***
2. ***{d.static.references.data[1]}***
3. ***{d.static.references.data[2]}***
4. ***{d.static.references.data[3]}***
5. ***{d.static.references.data[4]}***
6. ***{d.static.references.data[5]}***
7. ***{d.static.references.data[6]}***
8. ***{d.static.references.data[7]}***
9. ***{d.static.references.data[8]}***
10. ***{d.static.references.data[9]}***
11. ***{d.static.references.data[10]}***
12. ***{d.static.references.data[11]}***
13. ***{d.static.references.data[12]}***
14. ***{d.static.references.data[13]}***
15. ***{d.static.references.data[14]}***
16. ***{d.static.references.data[15]}***
17. ***{d.static.references.data[16]}***

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1. ***{d.static.references.data[17]}***
2. ***{d.static.references.data[18]}***
3. ***{d.static.references.data[19]}***
4. ***{d.static.references.data[20]}***
5. ***{d.static.references.data[21]}***
6. ***{d.static.references.data[22]}***
7. ***{d.static.references.data[23]}***
8. ***{d.static.references.data[24]}***
9. ***{d.static.references.data[25]}***
10. ***{d.static.references.data[26]}***
11. ***{d.static.references.data[27]}***

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*I\*

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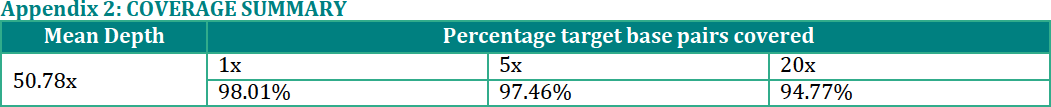
**Date Received: Indication** :



**07-Aug-2024**

## Appendix 1:SAMPLE DATA AND STATISTICS

|  |  |
| --- | --- |
| Title | Data |
|  |  |
|  |  |
|  |  |
|  |  |



**Appendix 3: COVERAGE OF ANALYZED GENES (Percentage of coding region covered)**

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| **Gene** | **Region Covered** | **Gene** | **Region covered** | **Gene** | **Region covered** |
|  |  |  |  |  |  |
|  |  |  |  |  |  |
|  |  |  |  |  |  |
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