**Dr eq**

# nucleome

LEARN I SERVEI DISCOVER SCIENCE

**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}

**Dr eq**

# nucleome

LEARN I SERVEI DISCOVER SCIENCE

**Name Sex/Age**

**Date Received: Indication** :

Phred Quality Score:

Genotype:

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

Depth:14

%

Allele DP

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

7

7

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):

**l**◄

**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]}



**Dr eq**

# nucleome

LEARN I SERVEI DISCOVER SCIENCE

**Name Sex/Age**

**Date Received: 07-Aug-2024 Indication** :

Methodology

{d.static.test\_methodology.data}

Limitations/Disclaimer:

d.static.limitation.data}

{d.static.disclaimer.data}

**Dr eq**

# nucleome

LEARN I SERVE I DISCOVER SCIENCE

**Name Sex/Age**

**Date Received: 07-Aug-2024 Indication** :

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]}

**Dr****eq**

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]}

**Dr eq**

*I\*

# nucleome

LEARN I SERVEI DISCOVER SCIENCE

**Name Sex/Age**

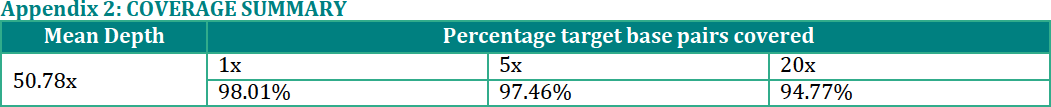
**Date Received: Indication** :



**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** |
|  |  |  |  |  |  |
|  |  |  |  |  |  |
|  |  |  |  |  |  |
|  |  |  |  |  |  |
|  |  |  |  |  |  |
|  |  |  |  |  |  |
|  |  |  |  |  |  |

----------·---·---·---EndOf Rpe o rt·---·---·-------------