Genome Variant Analysis Report

# Patient Information

Name: {d.patient.name}

Sex/Age: {d.patient.gender} / {d.patient.age}

Date Received: {d.patient.date\_received}

Indication: {d.indication}

# Whole Exome Sequencing on the Illumina Novaseq 6000 NGS Platform

Clinical Indication: {d.clinical\_indication}

## Interpretation

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| Gene & Transcript | Location | Variant | Zygosity/Inheritance | Clinical Significance |
| {d.variants[i].gene} | {d.variants[i].location} | {d.variants[i].variant} | {d.variants[i].zygosity} | {d.variants[i].clinical\_significance} |

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

## Interpretation

{d.interpretation}

## Variant Evidence

Chromosome: {d.chromosome}

Position: {d.Position}

Allele: {d.allele}

Depth (DP): {d.DP}

Percentage: {d.%}

Genotype: {d.heterozygous}

Phred Quality Score: {d.phredscore}

## Gene Impact

Transcript: {d.Transcript}

Protein: {d.protein}

Coding: {d.coding}

Based on above evidence, this variant {d.FLCN} is classified as Likely pathogenic variant.

## OMIM Phenotype

{d.OMIM Phenotype description}

## Case Specific Recommendations

{d.case\_specific\_recommendations}

{d.recommendations}

## Methodology

{d.methodology}

## Limitations/Disclaimer

{d.Limitations}