

Name :
Sex/Age :
Date Received :
Indication :

Whole Exome Sequencing on the Illumina Novaseq 6000 NGS Platform

Clinical Indication:

A 0.0 Year(s) old baby with gender - (not specified) born to parents with unknown consanguinity was referred for genetic evaluation. No clinical symptoms / phenotypes were mentioned.

*

Variant Interpretation & Clinical correlation:

Signature 1

Signature 2

Signature 3

Nucleome Address

Name :
Sex/Age :
Date Received :
Indication :

Variant Evidence			Gene Impact																
Chromosome: Position: NC_000017.11(GRCh38 Chr17):			RefSeq Gnese 110, NCBI																
<table border="1"> <thead> <tr> <th>Allele</th> <th>DP</th> <th>%</th> </tr> </thead> <tbody> <tr> <td> </td> <td> </td> <td> </td> </tr> <tr> <td> </td> <td> </td> <td> </td> </tr> </tbody> </table>			Allele	DP	%							<table border="1"> <tr> <td>Gene:</td> <td>Transcript:</td> </tr> <tr> <td>Effect:</td> <td>Protein:</td> </tr> <tr> <td>Exon:</td> <td>Coding:</td> </tr> </table>		Gene:	Transcript:	Effect:	Protein:	Exon:	Coding:
Allele	DP	%																	
Gene:	Transcript:																		
Effect:	Protein:																		
Exon:	Coding:																		
Genotype:			phred quality score:																

Based on the evidence, this variant is classified as likely to be variant

OMIM Phenotype:

Case Specific Recommendations:

Recommendations

Signature 1

Signature 2

Signature 3

Nucleome Address

Name :
Sex/Age :
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Methodology:

Limitations/Disclaimer:

Variant Classification as per ACMG guidelines:

References:

Signature 1

Signature 2

Signature 3

Nucleome Address

Name :
Sex/Age :
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Appendix 1: Sample Data and Statistics

Title	Data

Appendix 2: Coverage Summary

Mean Depth	Percentage target base pairs covered		

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

Gene	Region Covered	Gene	Region Covered	Gene	Region Covered