Dr Seq

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Name
Sex/Age Unknown/0
Date Received:
Indication Apr 03 2025

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.

Plausible cause was not identified.

Gene and Transcript	Location	Variant	Zygosity	OMIM Phenotype	Clinical Significance
chr16:2106263	chr16:2106263		Heterozygous	'POLYCYSTIC KIDNEY DISEASE 1 WITH OR WITHOUT POLYCYSTIC LIVER DISEASE'	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.

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

FLCN:c.1150_1160delGTCCAGTCAGC:

A Heterozygous variant NM_001009944.3:c.7531G>A, [NP_001009944.3:p.Ala2511Thr] in PKD1[MIM*601313] gene was identified by Whole Exome Proband-Only analysis. The Heterozygous variations in the PKD1 gene are known to cause Autosomal dominant POLYCYSTIC KIDNEY DISEASE 1 WITH OR WITHOUT POLYCYSTIC LIVER DISEASE [MIM#173900]. This variant is not present in publicly available databases like ['1000 Genomes', 'Genome Aggregation Database Exome (gnomAD_exome)', 'Genome Aggregation Database Genome (gnomAD_genome)', 'Inhouse exome database']. This variant is present in [['Exome Variant Server', '0.0']].

Reviewed by

[redacted]

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Sex/Age

Date Received: 07-Aug-2024

Indication :

Variant Evidence

Variant Evidence for 2400146295.targeted

GRCh37mm

Chromosome: PKD1

NC _000017 11 IGRCh38 Chrl7):

1

Position: PKD1

chr16:2106263

chr16:2106263

chr16:2106263

chr16:2106263

Gene Impact

Gene:

Transcript: PKD1

Complete. Reverse

Effect:

ProtP,n:

'POLYCYSTIC KIDNEY DISEASE 1 WITH OR WITHOUT POLYCYSTIC LIVER DISEASE'

Allele DP %

chr16:2106263

7

7

Depth:14

Coding:

Genotype: Heterozygous

Phred Quality Score:

Exon:

Based on the evidence, this variant Heterozygous is classified as likely to be 'POLYCYSTIC KIDNEY DISEASE 1 WITH OR WITHOUT POLYCYSTIC LIVER DISEASE' variant

OMIM Phenotype:

'POLYCYSTIC KIDNEY DISEASE 1 WITH OR WITHOUT POLYCYSTIC LIVER DISEASE'

Case Specific Recommendations:

- Genetic counselling is advised. For assistance in locating nearby genetic counseling services, please contact the laboratory [Ph.No. -------].
- Validation of the variant detected by Sanger sequencing is recommended.
- Targeted mutation analysis in the parents is recommended to document possible de novo status of the variant detected in the proband.

Reviewed by

[redacted]

[redacted]

[redacted]

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Name [redacted]

Sex/Age

Date Received: 07-Aug-2024

Indication :

Methodology

To be filled by organization based on the test methodology.

Limitations/Disclaimer

Absence of a plausible explanation for the reported phenotype by exome sequencing does not exclude a genetic basis of the patient's condition. Some types of genetic abnormalities, such as copy number changes, variants in non-coding regions, large insertions or deletions etc. may not be detectable in this exome analysis test. It is possible that the genomic region where a disease causing mutation exists in the proband was not captured in the current test and therefore was not detected. Additionally, it is possible that a particular genetic abnormality may not be recognized as the underlying cause of the genetic disorder due to incomplete scientific knowledge about the function of all genes in the human genome and the impact of variants in those genes. Only variants in genes associated with the medical condition, or thought to be clinically relevant potentially for the probands medical condition, are reported here.

Intronic and untranslated region variants are not assessed using this method.,The classification of variants may change over time.,Although all precautions have been taken during the test, the currently available data indicate that the chances of technical error are 2-3%.,For any further questions please contact the laboratory [Ph.No. -------].

[redacted]

[redacted]

[redacted]

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Variant Classification as per ACMG

Name	Description
Variant	A change in a gene. This could be disease causing (pathogenic) or not disease causing (benign).
Pathogenic	A disease causing variation in a gene which can explain the patients symptoms has been detected. This usually means that a suspected disorder for which testing had been requested has been confirmed.

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Appendix 1:SAMPLE DATA AND STATISTICS

Title	Data

Appendix 2: COVERAGE SUMMARY

Mean Depth	Percentage target base pairs covered		
50.78x	1x	5x	20x
	98.01%	97.46%	94.77%

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

Gene	Region Covered	Gene	Region covered	Gene	Region covered

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Reviewed by

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