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Name :
Sex/Age : Unknown/0 Year(s)
Date Received : Apr 03 2025
Indication :

Whole Exome Sequencing on the Illumina Novaseq 6000 NGS Platofrm

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:

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']].

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Variant Evidence	Gene Impact
Chromosome: Position: Allele / DP / % Heterozygous Genotype: Heterozygous phred quality score:	RefSeq Gnese 110, NCBI Gene: chr16:2106263 Transcript: chr16:2106263 Effect: 'POLYCYSTIC KIDNEY DISEASE 1 WITH OR WITHOUT POLYCYSTIC LIVER DISEASE' Protein: Exon: Coding:

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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

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.,Please note that the classification of variants may change over time if additional information becomes available.

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

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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. -------].

Variant Classification as per ACMG guidelines:

Name	Description

References:

McLaren, William, et al. The Ensembl Variant Effect Predictor. Genome Biology, vol. 17, no. 1, 6 June 2016, p. 122, doi:10.1186/s13059-016-0974-4.,1000 Genomes Project Consortium, et al. A Global Reference for Human Genetic Variation. Nature, vol. 526, no. 7571, 2015, pp. 68-74 doi:10.1038/nature15393.,Karczewski, Konrad J., et al. The ExAC Browser: Displaying Reference Data Information from over 60,000 Exomes. Nucleic Acids Research, vol. 45, no. D1, 2017, pp. D840-D845, doi:10.1093/nar/gkw971.,Chen, Shamil, et al. A Genomic Mutational Constraint Map Using Variation in 76,156 Human Genomes. Nature, vol. 625, no. 7993, 4 Jan. 2024, pp. 92-100.,Sherry, Stephen T., et al. dbSNP: The NCBI Database of Genetic Variation. Nucleic Acids Research, vol. 29, no. 1, 1 Jan. 2001, pp. 308-11.,Amberger, Joanna S., et al. OMIM.org: Online Mendelian Inheritance in Man (OMIMÂ®), an Online Catalog of Human Genes and Genetic Disorders. Nucleic Acids Research, vol. 43, no. D1, 28 Jan. 2015, pp. D789-98.,Landrum, Melissa J., et al. ClinVar: Public Archive of Interpretations of Clinically Relevant Variants. Nucleic Acids

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Appendix 1: Sample Data and Statistics

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

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Database Information:

1000 Genomes	Indigen	gnomAD_exome	gnom_genome	inhousedb	GME

DataBase Description: