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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.

Plausible cause was not identified.

*

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']].

Variant Evidence	Gene Impact
Chromosome: PKD1 Position: chr16:2106263	RefSeq Gnese 110, NCBI

Allele	DP	%

Gene:	Transcript:
chr16:2106263	chr16:2106263

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Heterozygous		

Genotype:
Heterozygous

Effect: 'POLYCYSTIC KIDNEY DISEASE 1 WITH OR WITHOUT POLYCYSTIC LIVER DISEASE'	Protein:
Exon:	Coding:

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'

Recommendations
• Genetic counselling is advised. For assistance in locating nearby genetic counseling services, please contact the laboratory [Ph.No. -------].

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- 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 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:

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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.

References:

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Appendix 1: Sample Data and Statistics

Appendix 2: Coverage Summary

Mean Depth	Percentage target base pairs covered

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

Gene	Region Covered	Gene	Region Covered	Gene	Region Covered

Database Information:
Database Information PKD1 : NM_001009944.3:c.7531G>A [NP_001009944.3:p.Ala2511Thr]

	1000 Genomes	Indigen	gnomAD_exome	gnom_genome	inhousedb	GME
Presence/ Absence	Absent	Present	Absent	Absent	Absent	Absent
MAF Vaues	MAF:NA	MAF:0.0	MAF:NA	MAF:NA	MAF:NA	MAF:NA

Additional Information:

Variant of unknown significance was identified.

Django Framework:

- Backend handles the logic for converting JSON data into a PDF format,Frontend provides a user-friendly interface for uploading JSON and DOCX template files.

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- Carbone API is used to authenticate requests to the Carbone service.
- Template Handling - handles docx template and json file with data.User can upload both

Workflow

Data Mapping: The JSON data is parsed and mapped onto the placeholders in the DOCX template.

- The mapping process in the project involves inserting JSON data into the placeholders within the DOCX template.
- When a user uploads a JSON file and a DOCX template, the Django backend parses the JSON data and identifies the corresponding placeholders in the DOCX file.

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- These placeholders are typically denoted by specific tags or markers (e.g.,).
- The system then replaces these placeholders with the actual data from the JSON file.

  For instance, if the JSON contains a patient's name, the system will find the placeholder for the patient's name in the template and replace it with the actual name from the JSON data.
- This process is repeated for all relevant fields, ensuring that the final DOCX document is fully populated with the provided data.
- Once the template is populated, it is sent to the Carbone API for conversion to a PDF document, which can then be downloaded by the user.
- This dynamic mapping ensures that the generated PDF accurately reflects the data provided in the JSON file, adhering to the format specified in the DOCX template.

- Users access the Django application through a web browser.
- The interface allows users to upload a DOCX template and a JSON file.
- Data Mapping
- The populated DOCX template is sent to the Carbone API for conversion to PDF.
- The Carbone API processes the request and returns the PDF document
- The generated PDF is made available for download through the user interface.