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REPORT

Name of Patient:

Referred by:

Sample received on: Apr 03 2025

Nature of sample: Blood

Test performed:

Age: 0Year(s)

Sex: Unknown

Date of report:

Ref. No.:

Clinical Summary

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.

Result

Plausible cause was not identified.

Additional Findings

Variant of unknown significance was identified.

Variant Information

Sample	Gene (Transcript) Disease (OMIM)	Location Inheritance	Variation	Zygosity	Classification
{#d.dynamic.variantInfo.data.rowData}					
{/d.dynamic.variantInfo.data.rowData}					

Database Information 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.

{/d.static.references.data}

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

{#d.dynamic.databaseInformation[0].data.rowData}

1000 Genomes GME	IndiGen	gnomAD_exome	gnomAD_genome	inhousedb
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Absent	Present	Absent	Absent	Absent	Absent
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{/d.dynamic.databaseInformation[0].data.rowData}

Interpretation

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']].

Recommendations

{#d.dynamic.recommendation.data}

-

Quality Metrics Information

PROBAND INFORMATION

Total Number of reads:

Number of reads on target:

Target size (bp):

Mean depth of coverage:

Quality threshold:

Terms and Conditions

[object Object]

Limitations

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.

Disclaimer

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. -----].

Notes

REFERENCES

{#d.static.references.data}

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