REPORT

Name of Patient: test_hg38

Referred by:

Sample received on: Apr 03 2025

Nature of sample: Blood

Test performed: Whole Exome

Age: 0Year(s)

Sex: Unknown

Date of report:

Ref. No.: test_hg38

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) | Location | Variation | Zygosity | Classification | Disease (OMIM) | Inheritance |

|--------|------------------|----------|-----------|----------|---------------|---------------|------------|

{#d.dynamic.variantInfo.data.rowData}

| test_hg38| PKD1| chr16:2106263| NM_001009944.3:c.7531G>A, NP_001009944.3:p.Ala2511Thr| Heterozygous| VUS| 'POLYCYSTIC KIDNEY DISEASE 1 WITH OR WITHOUT POLYCYSTIC LIVER DISEASE'| 'Autosomal dominant'|

{/d.dynamic.variantInfo.data.rowData}

Database Information

{#d.dynamic.databaseInformation}

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

| 1000 Genomes| IndiGen| gnomAD_exome| gnomAD_genome| inhousedb| GME|

|-------|-------|-------|-------|-------|-------|

{#d.dynamic.databaseInformation[$i].data.rowData}

| Absent| Present| Absent| Absent| Absent| Absent|

{/d.dynamic.databaseInformation[$i].data.rowData}

{/d.dynamic.databaseInformation}

Interpretation

Incidental or secondary findings (if any) that meet the ACMG guidelines can be provided upon request.

{/d.static.note.data}

Terms and Conditions

{#d.static.terms.data.rowData}

**Variant

**A change in a gene. This could be disease causing (pathogenic) or not disease causing (benign).

{/d.static.terms.data.rowData}

Limitations

Intronic and untranslated region variants are not assessed using this method.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}

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

{/d.dynamic.recommendation.data}

Test Methodology

To be filled by organization based on the test methodology.

Notes

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- Variant**: Description**: 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

{#d.static.disclaimer.data}

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