MAYO CLINIC
Mayo Medical Laboratories

Laboratory Service Report 1-800-533-1710

Patient Name	Patient ID	Age	Gender	Order #
SAMPLERECORD,ARPKM	SA00045521	45	F	SA00045521
Ordering Phys				**DOB** 06/10/1966
Client Order # SA00045521	**Account Information**			**Report Notes**
Collected 04/29/2012	C7028846-DLMP ROCHESTER 3050 SUPERIOR DRIVE ROCHESTER,MN 55901			
Printed 07/17/2012 17:37				

Test	Flag	Results	Unit	Reference Value	Perform Site*
ARPKD Known Mutation			REPORTED 07/13/2012 09:50		
Specimen		Blood			MCR
Specimen ID		1038127			MCR
Order Date		08 May 2012 07:41			MCR
Reason For Referral					MCR

Family history of autosomal recessive polycystic kidney disease (ARPKD). Test for the presence of familial alterations in the PKHD1 gene.

Method MCR

DNA sequence analysis was used to test for the presence of the p.T36M (c.107C>T) and p.R1624W (c.4870C>T) alterations in exons 3 and 32 respectively, in the PKHD1 gene. Analysis for these specific alterations was performed because they were identified in a family member. Mutation nomenclature is based on GenBank accession number NM_138694.3.

Result MCR

The following heterozygous sequence changes were detected:
Exon: 3
DNA change: c.107C>T
Amino Acid change: p.T36M (Thr36Met)
Classification: DELETERIOUS

AND

Exon: 32
DNA change: c.4870C>T
Amino Acid change: p.R1624W (Arg1624Trp)
Classification: DELETERIOUS

Interpretation MCR

The p.T36M and p.R1624W alterations are known deleterious mutations.

This result is consistent with a diagnosis of ARPKD.

Since mutations have been identified in the PKHD1 gene, genetic testing of at risk family members is possible.

A genetic consultation may be of benefit.

Unless reported or predicted to cause disease, alterations found deep in the intron or alterations that do not result in an amino acid substitution are not reported. These and common polymorphisms identified for this patient are available upon request.

Performing Site Legend on Last Page of Report

* Report times for Mayo performed tests are CST/CDT

TEST SITE

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CAUTIONS:
Test results should be interpreted in context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Rare polymorphisms exist that could lead to false negative or positive results. If results obtained do not match the clinical findings, additional testing should be considered.

Bone marrow transplants from allogenic donors will interfere with testing. Call Mayo Medical Laboratories for instructions for testing patients who have received a bone marrow transplant.

Laboratory developed test.

Reviewed By: MCR
Melody Elizabeth Kimball

Release Date 13 Jul 2012 09:48 MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:

Patient Name	Collection Date and Time	Report Status
SAMPLERECORD,ARPKM	04/29/2012	Final
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TEST SITE