

Patient Name SAMPLEREP,IVD	Patient ID SA00046801	Age 45	Gender F	Order # SA00046801
Ordering Phys			DOB 06/10/1966	
Client Order # SA00046801	Account Information C7028846-DLMP Rochester 3050 Superior Drive Rochester, MN 55901			Report Notes
Collected 05/24/2012				
Printed 05/30/2013 16:03				

Test	Flag	Results	Unit	Reference Value	Perform Site*
IVD Mutation Analysis (A282V)				REPORTED 07/13/2012 10:27	
Specimen		Blood			MCR
Specimen ID		1038234			MCR
Order Date		29 May 2012 08:47			MCR
Reason For Referral		Patient reported to have features suggestive of isovaleric acidemia. Test for the presence of the A282V mutation within the isovaleryl-CoA dehydrogenase (IVD) gene.			MCR
Method		A PCR-based assay was used to examine for the presence of the A282V mutation within the isovaleryl-CoA dehydrogenase (IVD) gene.			MCR
Result		The A282V (Ala282Val) mutation within the isovaleryl-CoA dehydrogenase (IVD) gene was not detected.			MCR
Interpretation		This result does not rule out the diagnosis of isovaleric acidemia. Several other disease causing mutations have been described in the IVD gene. Therefore, absence of this mutation does not eliminate the possibility of a diagnosis of isovaleric acidemia.			MCR
		Results should be interpreted in light of clinical history and other laboratory analyses (i.e. plasma acylcarnitine, urine organic acid, and urine acylglycine).			
		CAUTIONS: 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.			
		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.			
		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:		Melody Elizabeth Kimball			MCR

Performing Site Legend on Last Page of Report

Patient Name SAMPLEREP,IVD	Collection Date and Time 05/24/2012	Report Status Final
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* Report times for Mayo performed tests are CST/CDT

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Test	Flag	Results	Unit	Reference Value	Perform Site*
Release Date		13 Jul 2012 10:25			MCR

* Performing Site:

MCR	Mayo Clinic Laboratories - Rochester Main Campus 200 First St SW Rochester, MN 55905	Lab Director:
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Patient Name SAMPLEREP,IVD	Collection Date and Time 05/24/2012	Report Status Final
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