

<b>Patient Name</b> TESTING,84209	<b>Patient ID</b>	<b>Age</b>	<b>Gender</b>	<b>Order #</b> W3088154
<b>Ordering Phys</b>		<b>DOB</b>		
<b>Client Order #</b> W3088154	<b>Account Information</b> C7999998-STUSTEST 200 FIRST STREET SW ROCHESTER, MN 55901	<b>Report Notes</b>		
<b>Collected</b> 10/30/2009 06:00				
<b>Printed</b> 11/02/2009 13:38	(507)266-5730			

Test	Flag	Results	Unit	Reference Value	Perform Site*
<b>Factor IX Gene Mutation Screening</b>			REPORTED	11/02/2009 07:59	
F IX Mut Screen Reason for Referral		No charge.			MCR
Factor IX Mutation Screen Method		Regions analyzed include the promoter region, coding regions (exons 1 through 8, including splice junctions) and a portion of the 3' untranslated region. This test is a direct mutation analysis of leukocyte genomic DNA performed by polymerase chain reaction, followed by sequencing utilizing fluorescent ABI technology, Foster City, CA.			MCR
Factor IX Mutation Screen Result		Results: A homozygous nucleotide change C20413T (point mutation) in exon 6 of FIX gene, resulting in a missense mutation amino acid change Arg145Cys was found in Testing Patient.			MCR
Factor IX Mutation Screen Interp		Interpretation: Mutations: The Arg 145 Cys mutation has been demonstrated in patients with moderate congenital hemophilia B (factor IX deficiency). Results therefore are consistent with a diagnosis of hemophilia B in Testing Patient. This assay will not detect mutations known to cause congenital hemophilia B outside the regions described in methods section and cannot exclude the possibility of somatic mosaicism which has been reported in congenital hemophilia B. Genetic consultation may be of benefit for this individual and/or family to further discuss the implications of these findings.			MCR
Factor IX Mut Screen Reviewed By		R.K. Pruthi, MD			MCR
Please note change in reporting and methodology effective 06/19/2008.					
<b>Factor IX Mut Scrn Gene Sequencing</b>			REPORTED	11/02/2009 07:59	
Sequencing		Performed			MCR

\* Performing Site:

MCR	Mayo Clinic Dpt of Lab Med & Pathology 200 First St SW Rochester, MN 55905	Lab Director: Franklin R. Cockerill, III, M.D.
-----	---	--

<b>Patient Name</b> TESTING,84209	<b>Collection Date and Time</b> 10/30/2009 06:00	<b>Report Status</b> Final
Page 1 of 1		** End of Report **

\* Report times for Mayo performed tests are CST/CDT