

Result Summary-Positive\n\n Result â□The following heterozygous alteration was identified:  
Amino Acid change: p.Q188R (Gln188Arg). DNA change: c.563A>G (g.34648167),  
Classification: PATHOGENIC \n\n Interpretation - Biochemical and molecular test results are in  
agreement. The observed GALT enzyme activity in red blood cells (12.2 nmol\*\*mg Hb) and the  
presence of a single copy of p.Q188R suggest that this individual is a carrier of classic  
galactosemia. This individual should not be at risk for developing symptoms related to this  
disorder; however, he or she may be at risk for having offspring with galactosemia. If appropriate,  
enzymatic and molecular studies for this individual's reproductive partner are recommended to  
further clarify this risk. \n\n Method - A multiplex PCR-based assay was used to test for the  
presence of the following mutations in the GALT gene.