

Result Summary-Positive
Result : The following heterozygous alteration was identified:
Amino Acid change: p.Q188R (Gln188Arg). DNA change: c.563A>G (g.34648167),
Classification: PATHOGENIC
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.
Method - A multiplex PCR-based assay was used to test for the presence of the following mutations in the GALT gene.