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## Standardization of PCR based assays to identify common deletions in the GALT Gene

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Classical Galactosemia is an autosomal recessive deficiency of the enzyme galactose-1-phosphate uridyltransferase (GALT) resulting in elevated levels of galactose (1). Untreated infants presents with life-threatening complications that include feeding problems, failure to thrive, liver damage, cataracts, bleeding, sepsis and neonatal death (2). Its incidence is 1/47,000 in Caucasian and 1/20,000 in African-American populations. A galactose-free diet improves symptoms and signs. Several known mutations (i.e. Q188R, S135L, K285N) have been associated with a classical galactosemia phenotype. Two different GALT gene deletions have also been reported mainly in the Ashkenazi Jewish population.

The aim of this study is to standardize and validate the two different ~ 5 Kb deletions characterized by Coffee et al. and Berry et al. These assays will be added to the panel of GALT gene mutations tested by the New York State Newborn Screening program.

References:

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3. Berry, G.T., Leslie, N., Reynolds, R., Yager, C.T., & Segal, Stanton (2001). Evidence for Alternate Galactose Oxidation in a Patient with Deletion of the Galactose-1-Phosphate Uridyltransferase Gene. *Molecular Genetics and Metabolism* . 72, 316-321.