NEWBORN SCREENING FOR GALACTOSEMIA: DUARTE VARIANT BIOCHEMICAL PHENOTYPE AND CLINICAL OUTCOMES.

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** Purpose:** Newborn screening (NBS) for galactosemia was established to identify significantly affected infants at high risk for liver disease, sepsis and growth failure. Current screening by galactose-1-phosphate uridyltransferase (GALT) enzyme analysis in newborn blood spots identifies a large number of patients with Duarte galactosemia, a mild variant form of galactosemia. Currently no standard of care for the treatment of DG galactosemia exists, although many centers completely restrict dietary lactose until age 1 year. This study identifies outcomes in patients with DG galactosemia after identification by NBS, dietary lactose restriction for 1 year and 3-5 years follow-up.

**Methods:** Clinical and biochemical parameters were summarized in the management of 57 patients diagnosed with Duarte galactosemia identified by NBS in Louisiana between 2/15/2003 and 10/30/2008. Patients in the study were followed to a median age of 53 months (age range 17-82 months). **Results:** Patients had a mean GALT activity of 40% of normal (range = 7.4-77.5%). All patients were confirmed by GALT mutation analysis and/or isozyme analysis. After one year of lactose restriction, all patients who were examined (42/57 = 74%) had normal levels of red blood cell galactose-1-phosphate, a marker for treatment efficacy. Physical development data of patients (46/57 = 81%) at an average age of 12.8 months (range = 1-36 months) showed normal growth on three standard growth parameters with Z-scores for height-for-age (0.10 ± 1.13), weight-for-age (0.32 ± 1.66), and head circumference-for-age (0.38 ± 1.21) essentially normal. Liver function studies on a subset of patients (22/57 = 39%) revealed mild AST elevation in only 2 patients and ALT elevation in one patient. None of the patients who received ophthalmologic evaluation (12/57 = 21%) showed development of cataracts. No patients had signs of developmental delay or had been referred for developmental abnormalities. **Conclusions:** NBS for galactosemia identifies a large proportion of children with Duarte variant galactosemia, for which no standard of care currently exists. This study reports that after galactose restriction for one year, all examined patients had normal biochemical parameters and growth development within normal range.

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