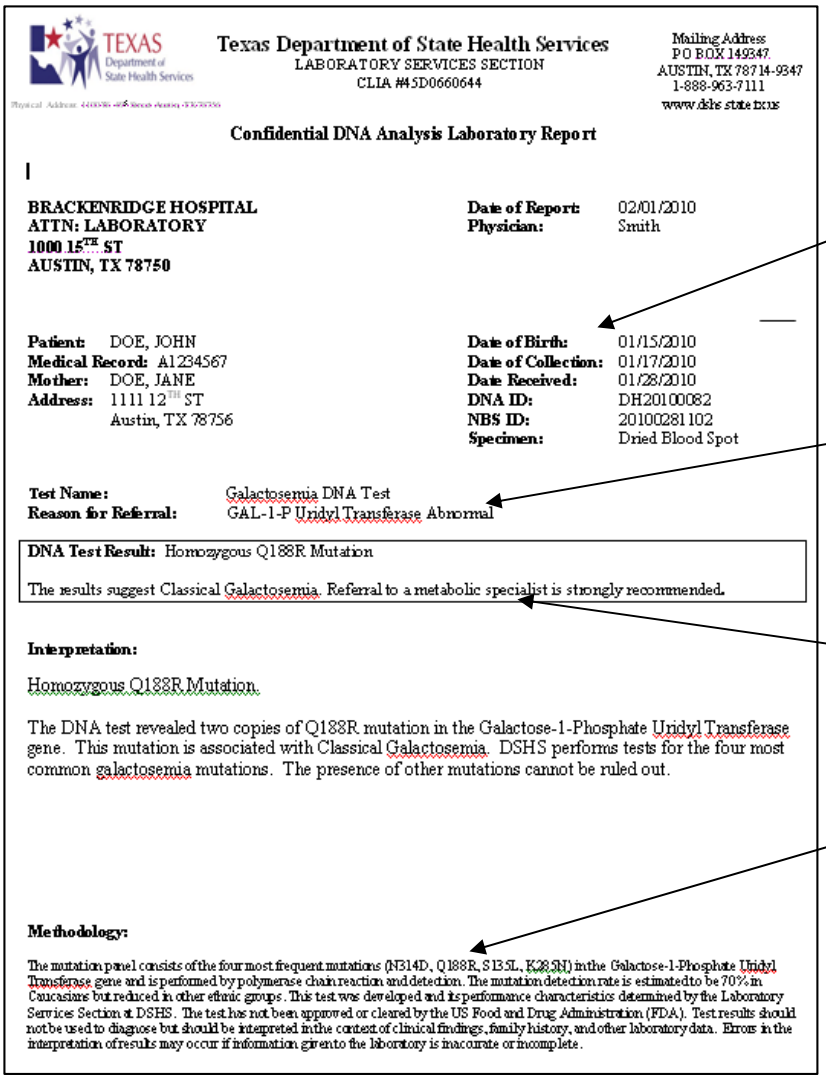
Notice

Changes in Galactosemia DNA Report

The DSHS Newborn DNA Analysis laboratory implemented a molecular second-tier test for galactosemia in May 2007 as a part of the newborn screening program. The current testing panel targets four common galactosemia mutations. The DNA reports are now mailed directly to the providers. The format of the Galactosemia DNA report has been modified to make it easier to read and understand. Please see below for highlights of the changes.



TEXAS Department of State Health Services

Texas Department of State Health Services
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Confidential DNA Analysis Laboratory Report

BRACKENRIDGE HOSPITAL
ATTN: LABORATORY
1000 15TH ST
AUSTIN, TX 78750

Date of Report: 02/01/2010
Physician: Smith

Patient: DOE, JOHN
Medical Record: A1234567
Mother: DOE, JANE
Address: 1111 12TH ST
Austin, TX 78756

Date of Birth: 01/15/2010
Date of Collection: 01/17/2010
Date Received: 01/28/2010
DNA ID: DH20100082
NBS ID: 20100281102
Specimen: Dried Blood Spot

Test Name: Galactosemia DNA Test
Reason for Referral: GAL-1-P Uridyl Transferase Abnormal

DNA Test Result: Homozygous Q188R Mutation

The results suggest Classical Galactosemia. Referral to a metabolic specialist is strongly recommended.

Interpretation:

Homozygous Q188R Mutation.

The DNA test revealed two copies of Q188R mutation in the Galactose-1-Phosphate Uridyl Transferase gene. This mutation is associated with Classical Galactosemia. DSHS performs tests for the four most common galactosemia mutations. The presence of other mutations cannot be ruled out.

Methodology:

The mutation panel consists of the four most frequent mutations (N314D, Q188R, S135L, K285N) in the Galactose-1-Phosphate Uridyl Transferase gene and is performed by polymerase chain reaction and detection. The mutation detection rate is estimated to be 70% in Caucasians but reduced in other ethnic groups. This test was developed and its performance characteristics determined by the Laboratory Services Section at DSHS. The test has not been approved or cleared by the US Food and Drug Administration (FDA). Test results should not be used to diagnose but should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in the interpretation of results may occur if information given to the laboratory is inaccurate or incomplete.

Includes Pertinent Demographic Information

Provides Reason for Referral

Easier to Locate DNA Test Result

Addition of a detailed methodology section, including a listing of targeted mutations and estimated mutation detection rates.

Any questions? Contact the Newborn Screening Laboratory
1-800-252-8023 ext. 7158