Title: Duarte Variant Galactosemia *GeneReview*, Biochemical Tests of Historical Interest

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Note: The following information is provided by the authors and has not been reviewed by *GeneReviews* staff.

Biochemical Tests of Historical Interest

Two additional biochemical tests, listed below, have been used historically to assist in diagnosis of Duarte variant galactosemia:

- Isoelectric focusing of native GALT isozymes from erythrocytes followed by a GALT enzyme activity overlay stain reveals a shifted pattern of GALT bands in Duarte variant galactosemia samples relative to controls [Beutler et al 1965]. This shift reflects charge alterations of the GALT protein due to substitution of a negatively charged aspartate (D) in place of asparagine (N) at residue 314 [Fridovich-Keil et al 1995].
- Whole body oxidation of ^{13}C-labeled galactose results have been reported for one six year old and one adult with Duarte variant galactosemia [Berry et al 1995]; in both cases the *in vivo* oxidation of galactose appeared normal.

Note: (1) *GALT* sequence analysis combined with standard testing of red blood cell GALT enzyme activity has effectively replaced the need for isoelectric focusing. (2) Although whole body oxidation of ^{13}C-labeled galactose may still be used in some cases to help determine an individual's whole body capacity for galactose metabolism, other alternatives (e.g., urinary galactitol) are also available and may be more cost effective.

References

Berry GT, Nissim I, Mazur AT, Elsas LJ, Singh RH, Klein PD, Gibson JB, Lin Z, Segal S. In vivo oxidation of [13C]galactose in patients with galactose-1-phosphate uridyltransferase deficiency. Biochem Mol Med. 1995:56:158-65.

Beutler E, Baluda ML, Sturgeon P, Day RW. A new genetic abnormality resulting in galactose-1-phosphate uridyltransferase deficiency. Lancet. 1965;1:353-4.

Fridovich-Keil JL, Quimby BB, Wells L, Mazur LA, Elsevier JP. Characterization of the N314D allele of human galactose-1-phosphate uridylyltransferase using a yeast expression system. Biochem Mol Med. 1995:56:121-30.