

Product datasheet for **TP720236L**

GALE (NM_000403) Human Recombinant Protein

Product data:

Product Type:	Recombinant Proteins
Description:	Recombinant protein of human UDP-galactose-4-epimerase (GALE), transcript variant 1
Species:	Human
Expression Host:	E. coli
Expression cDNA Clone or AA Sequence:	Met1-Ala348
Tag:	N-His
Predicted MW:	40.4 kDa
Concentration:	lot specific
Purity:	>95% as determined by SDS-PAGE and Coomassie blue staining
Buffer:	Provided lyophilized from a 0.2 µm filtered solution of 20 mM Tris-HCl, 150 mM NaCl
Endotoxin:	< 0.1 EU per µg protein as determined by LAL test
Storage:	Store at -80°C.
Stability:	Stable for at least 3 months from date of receipt under proper storage and handling conditions.
RefSeq:	NP_000394
Locus ID:	2582
UniProt ID:	Q14376 , A0A384NL38
Cytogenetics:	1p36.11
Synonyms:	SDR1E1



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Summary:

This gene encodes UDP-galactose-4-epimerase which catalyzes two distinct but analogous reactions: the epimerization of UDP-glucose to UDP-galactose, and the epimerization of UDP-N-acetylglucosamine to UDP-N-acetylgalactosamine. The bifunctional nature of the enzyme has the important metabolic consequence that mutant cells (or individuals) are dependent not only on exogenous galactose, but also on exogenous N-acetylgalactosamine as a necessary precursor for the synthesis of glycoproteins and glycolipids. Mutations in this gene result in epimerase-deficiency galactosemia, also referred to as galactosemia type 3, a disease characterized by liver damage, early-onset cataracts, deafness and cognitive disability, with symptoms ranging from mild ('peripheral' form) to severe ('generalized' form). Multiple alternatively spliced transcripts encoding the same protein have been identified. [provided by RefSeq, Jul 2008]

Protein Families:

Druggable Genome

Protein Pathways:

Amino sugar and nucleotide sugar metabolism, Galactose metabolism, Metabolic pathways

Product images: