MAYO CLINIC LABORATORIES

Test Definition: TALDO

Polyols, QN, U

Overview

Useful For

Diagnosis of transaldolase deficiency or ribose-5-phosphate isomerase deficiency

Genetics Test Information

This is a screening test for transaldolase deficiency or ribose-5-phosphate isomerase deficiency.

Highlights

Transaldolase (TALDO) deficiency and ribose-5-phosphate (RPI) deficiency are 2 recently described multisystem disorders of the pentose phosphate pathway.
Polyols analysis in urine is the method of choice for detecting TALDO deficiency and RPI deficiency.

Method Name

Gas Chromatography-Mass Spectrometry (GC-MS)

NY State Available

Yes

Specimen

Specimen Type

Urine

Necessary Information

Patient's age is required.

Specimen Required

Supplies: Urine Tubes, 10 mL (T068)
Specimen Volume: 2 mL
Collection Instructions:
1. Collect a random urine specimen.
2. No preservative.

Forms

If not ordering electronically, complete, print, and send a Biochemical Genetics Test Request (T798) with the specimen.

Reject Due To

All specimens will be evaluated at Mayo Clinic Laboratories for test suitability.

Specimen Minimum Volume

1 mL

Specimen Stability Information

Specimen Type	Temperature	Time	Special Container
Urine	Refrigerated (preferred)	28 days	
	Frozen	28 days	

Clinical & Interpretive

Clinical Information

Polyols are sugar alcohols that have been identified in blood, urine, and cerebrospinal fluid. Characteristic patterns of abnormal polyols may suggest a disorder of the pentose phosphate pathway (PPP) including transaldolase (TALDO) deficiency and ribose-5-phosphate isomerase (RPI) deficiency. The PPP is involved in carbohydrate metabolism and is present in the cytosol of all cells. Two specific functions of the PPP are the production of nicotinamide adenine dinucleotide phosphate and the synthesis of ribose-5-phosphate, a molecule necessary for nucleotide and nucleic acid synthesis. Both TALDO and RPI deficiency, which have multisystem involvement are recently described disorders of this pathway.

TALDO deficiency is an autosomal recessive disorder caused by a reduction of the enzyme transaldolase. Clinical manifestations are characterized by severe neonatal liver failure, coagulopathy, low serum protein, hypoglycemia, high ammonia, progressive myocardial hypertrophy, and abnormal lactate dehydrogenase with remarkably normal or low transaminases.

Patients may present in the antenatal period with maternal HELLP syndrome (hemolysis, elevated liver enzymes, low platelets), hydrops fetalis and oligohydramnios, dysmorphic features, cutis laxa, and hypertrichosis. The clinical course is variable, but acute liver failure with normal transaminases is a common finding. Initially, hepatomegaly is absent, but the spleen may be enlarged. Later, hepatomegaly with liver cirrhosis and mild kidney failure occur.

RPI deficiency is an autosomal recessive disorder caused by a deficiency of the enzyme ribose-5-phosphate isomerase. Clinical manifestations include neurological deficits such as slow progressing leukoencephalopathy and neuropathy. Additionally, spasticity, ataxia, epilepsy, regression, and delayed psychomotor development have been described.

Polyols analysis in urine is the method of choice for the biochemical diagnosis of TALDO and RPI deficiency. Abnormal results should be followed with either enzymatic or molecular genetic analysis.

Reference Values

ERYTHRITOL

< or =11 months: <220 mmol/mol creatinine

1-3 years: <267 mmol/mol creatinine

4-17 years: <171 mmol/mol creatinine

>or =18 years: <99 mmol/mol creatinine

ARABINITOL

< or =11 months: <140 mmol/mol creatinine

1-3 years: <149 mmol/mol creatinine

4-17 years: <97 mmol/mol creatinine

>or =18 years: <51 mmol/mol creatinine

RIBITOL

< or =11 months: <31 mmol/mol creatinine

1-3 years: <31 mmol/mol creatinine

4-17 years: <17 mmol/mol creatinine

>or =18 years: <11 mmol/mol creatinine

SEDOHEPTULOSE
< or =11 months: <76 mmol/mol creatinine
1-3 years: <27 mmol/mol creatinine
4-17 years: <28 mmol/mol creatinine
>or =18 years: <22 mmol/mol creatinine

Interpretation

An interpretive report will be provided.
All profiles are reviewed by the laboratory director and interpretation is based on pattern recognition. A detailed interpretation is given, including an overview of the results and of their significance, a correlation to available clinical information, elements of differential diagnosis, recommendations for additional biochemical testing and in vitro confirmatory studies (enzyme assay, molecular analysis), name and phone number of key contacts who may provide these studies at Mayo or elsewhere, and a phone number to reach one of the laboratory directors in case the referring physician has additional questions.

Cautions

A positive test result is diagnostic of transaldolase deficiency or ribose-5-phosphate isomerase deficiency; however, it is strongly recommended to follow-up with molecular analysis.

Clinical Reference

1. Wamelink MC, Valayannopoulos V, Jakobs C. Ribose-5-phosphate isomerase deficiency and transaldolase deficiency. In: Valle DL, Antonarakis S, Ballabio A, Beaudet AL, Mitchell GA. eds. The Online Metabolic and Molecular Bases of Inherited Disease. McGraw Hill; 2019. Accessed July 16, 2021. Available at www.ommbid.com
2. Eyaid W, Al Harbi T, Anazi S, et al: Transaldolase deficiency: report of 12 new cases and further delineation of the phenotype. J Inherit Metab Dis. 2013;36:997-1004
3. Huck JH, Verhoeven NM, Struys EA, et al: Ribose-5-phosphate isomerase deficiency: new inborn error in the pentose phosphate pathway associated with a slowly progressive leukoencephalopathy. Am J Hum Genet. 2004;74:745-751
4. Wamelink MM, Struys EA, Jakobs C: The biochemistry, metabolism and inherited defects of the pentose phosphate pathway: a review. J Inherit Metab Dis. 2008;31:703-717

Performance

Method Description

Urine specimens are spiked with a mixture of labeled internal standards, allowed to equilibrate, and evaporated. The dry residue is derivatized to form trimethylsilyl esters then extracted with hexane. Specimens are analyzed by gas chromatography/mass spectrometry, selected ion monitoring using ammonia chemical ionization and a stable isotope dilution method.(Jansen G, Muskiet F, Schierbeek H, et al: Capillary gas chromatography profiling of urinary, plasma, and erythrocyte sugars and polyols as their trimethylsilyl derivatives, preceded by a simple and rapid prepurification method. Clin Chim Acta 1986;157:277-294, Kaur P, Wamelink MMC, van der Knaap MS, et al: Confirmation of a rare genetic leukoencephalopathy due to a novel bi-allelic variant in RPIA. Eur J Med Genet. 2019 Aug;62(8):103708. doi: 10.1016/j.ejmg.2019.103708)

PDF Report

No

Specimen Retention Time
3 months

Performing Laboratory Location
Rochester

Fees & Codes

Test Classification
This test was developed, and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the US Food and Drug Administration.

CPT Code Information
82542