Reporting Title: GALT Gene, Full Gene Analysis
Performing Location: Rochester

Specimen Requirements:

Forms:
1. Molecular Genetics-Biochemical Disorders Patient Information Sheet (Supply T527) in Special Instructions
2. New York Clients-Informed consent is required. Please document on the request form or electronic order that a copy is on file. An Informed Consent for Genetic Testing (Supply T576) is available in Special Instructions.
3. If not ordering electronically, submit a Molecular Genetics Request Form (Supply T245) with the specimen.

Specimen must arrive within 96 hours of draw.

Submit only 1 of the following specimens:

Specimen Type: Whole blood
Container/Tube:
Preferred: Lavender top (EDTA) or yellow top (ACD)
Acceptable: Any anticoagulant
Specimen Volume: 3 mL
Collection Instructions:
1. Invert several times to mix blood.
2. Send specimen in original tube.
Specimen Stability Information: Ambient (preferred)/Refrigerated

Specimen Type: Blood spot
Container/Tube:
Preferred: Collection card (Whatman Protein Saver 903 Paper)
Acceptable: Ahlstrom 226 filter paper, or Supplemental Newborn Screening Card (Supply T493)
Specimen Volume: 2 to 5 Blood Spots on collection card (Whatman Protein Saver 903 Paper; Ahlstrom 226 filter paper; or Supplemental Newborn Screening Card, Supply T493)
Collection Instructions:
1. An alternative blood collection option for a patient >1 year of age is finger stick.
2. Let blood dry on the filter paper at ambient temperature in a horizontal position for 3 hours.
3. Do not expose specimen to heat or direct sunlight.
4. Do not stack wet specimens.
5. Keep specimen dry
Specimen Stability Information: Ambient (preferred)/Refrigerated

Specimen Type	Temperature	Time
Varies	Ambient (preferred)	
	Frozen	
	Refrigerated	

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
27413	Specimen	Alphanumeric		N/A
27414	Specimen ID	Alphanumeric		N/A
27415	Source	Alphanumeric		31208-2
27416	Order Date	Alphanumeric		N/A
27417	Reason For Referral	Alphanumeric		42349-1
27418	Method	Alphanumeric		49549-9
27419	Result	Alphanumeric		42318-6
27420	Interpretation	Alphanumeric		69047-9
27421	Amendment	Alphanumeric		42318-6
27422	Reviewed By	Alphanumeric		N/A
27423	Release Date	Alphanumeric		N/A

CPT Code: 1 × 81406

Reference Values:

An interpretive report will be provided.