

Test Definition: GALCS

Krabbe Disease, Full Gene Analysis and Large (30 kb) Deletion, PCR

Reporting Title: Krabbe Disease, Full Gene Analysis

Performing Location: Rochester

Specimen Requirements:

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.

Specimen must arrive within 96 hours of collection.

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:

Invert several times to mix blood.
 Send specimen in original tube.

Specimen Stability Information: Ambient (preferred)/Refrigerated

Specimen Type: Cultured fibroblasts Container/Tube: T-75 or T-25 flask

Specimen Volume: 1 full T-75 or 2 full T-25 flasks

Specimen Stability Information: Ambient (preferred)/Refrigerated <24 hours

Specimen Type: Skin biopsy

Container/Tube: Sterile container with any standard cell culture media (eg, minimal essential media, RPMI 1640). The solution should be supplemented with 1% penicillin and streptomycin. Tubes can be supplied upon request (Eagle's minimum essential medium with 1% penicillin and streptomycin [Supply T115]).

Specimen Volume: 4-mm punch

Specimen Stability Information: Refrigerated (preferred)/Ambient

Acceptable:

Specimen Type: Blood spot

Container/Tube: Whatman Protein Saver 903 Paper

Specimen Volume: 5 blood spots

Collection Instructions:

- 1. Let blood dry on the filter paper at ambient temperature in a horizontal position for 3 hours.
- 2. Do not expose specimen to heat or direct sunlight.
- 3. Do not stack wet specimens.
- 4. Keep specimen dry.

Specimen Stability Information: Ambient (preferred)/Refrigerated

Specimen Type	Temperature	Time
Varies	Varies	

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Ask at Order Entry (AOE) Questions:

Test ID	Question ID	Description	Туре	Reportable
FBC	CG030	Source: • Autopsy • Chorionic Villi • Products of Conception or Stillbirth	Answer List	No
FBC	CG033	Reason for Referral	Plain Text	No

Result Codes:

Result ID	Reporting Name	Туре	Unit	LOINC®
33569	Release Date	Alphanumeric		
33570	Specimen ID	Alphanumeric		
33571	Source	Alphanumeric		
33572	Order Date	Alphanumeric		
33573	Reason For Referral	Alphanumeric		
33574	Method	Alphanumeric		
33575	Result	Alphanumeric		
33576	Interpretation	Alphanumeric		
33577	Amendment	Alphanumeric		
33578	Reviewed By	Alphanumeric		
33579 Release Date		Alphanumeric		

CPT Code Information:

Krabbe Disease, Full Gene Analysis

83891-Isolation or extraction of highly purified nucleic acid

83894-Separation by gel electrophoresis

83898 x 20-Amplification, target, each nucleic acid sequence

83909 x 38-Separation and identification by high-resolution technique

83912-Interpretation and report

Fibroblast Culture for Genetic Testing

88233-Tissue culture, skin or solid tissue biopsy (if appropriate)

88240-Cryopreservation (if appropriate)

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Reflex Tests:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
FBC	Fibroblast Culture for Genetic Test		Profile	No	Yes

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Туре	Unit	LOINC®
FBC	16167	Specimen	Alphanumeric		
FBC	16337	Specimen ID	Alphanumeric		
FBC	CG030	Source	Alphanumeric		
FBC	16168	Order Date	Alphanumeric		
FBC	CG033	Reason For Referral	Alphanumeric		
FBC	16172	Method	Alphanumeric		
FBC	16175	Interpretation	Alphanumeric		
FBC	16176	Amendment	Alphanumeric		
FBC	16177	Consultant	Alphanumeric		
FBC	16178	Report Date	Alphanumeric		

Reference Values:

An interpretive report will be provided.