Reporting Title: Krabbe Disease, Known Mutation
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.

Specimen must arrive within 96 hours of collection.

Submit only 1 of the following specimens:

Preferred:
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: 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

All prenatal specimens must be accompanied by a maternal blood specimen. Order MCC/88636 Maternal Cell Contamination, Molecular Analysis on both the prenatal and maternal specimens. Due to the complexity of prenatal testing, consultation with the laboratory is required for all prenatal testing.

Specimen Type: Amniotic fluid
Container/Tube: Amniotic fluid container
Specimen Volume: 20 mL
Specimen Stability Information: Refrigerated (preferred)/Ambient

Specimen Type: Chorionic villi
Container/Tube: 15-mL tube containing 15-mL of transport media
Specimen Volume: 20 mg
Specimen Stability Information: Refrigerated

Acceptable:
Specimen Type: Confluent cultured cells
Container/Tube: T-25 flask
Specimen Volume: 2 flasks
Collection Instructions: Submit confluent cultured cells from another laboratory.
Specimen Stability Information: Ambient (preferred)/Refrigerated

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	

Ask at Order Entry (AOE) Questions:

Test ID	Question ID	Description	Type	Reportable
FBC	CG030	Source: • Autopsy • Chorionic Villi • Products of Conception or Stillbirth	Answer List	No
FBC	CG033	Reason for Referral	Plain Text	No
AFC	CG132	Reason For Referral	Plain Text	No

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
33580	Specimen	Alphanumeric		
33581	Specimen ID	Alphanumeric		
33582	Source	Alphanumeric		
33583	Order Date	Alphanumeric		

Result ID	Reporting Name	Type	Unit	LOINC®
33584	Reason For Referral	Alphanumeric		
33585	Method	Alphanumeric		
33586	Result	Alphanumeric		
33587	Interpretation	Alphanumeric		
33588	Amendment	Alphanumeric		
33589	Reviewed By	Alphanumeric		
33590	Release Date	Alphanumeric		

CPT Code Information:

Krabbe Disease, Known Mutation
83891-Isolation or extraction of highly purified nucleic acid
83912-Interpretation and report
DNA Sequence, Follow-up Analysis
83892-Enzymatic digestion (if appropriate)
83894-Separation by gel electrophoresis (if appropriate)
83898-Amplification, target, each nucleic acid sequence (if appropriate)
83909 x 2-Separation and identification by high-resolution technique (if appropriate

Follow Up PCR
83894-Separation by gel electrophoresis (if appropriate)
83898-Amplification, target, each nucleic acid sequence (if appropriate)

Amniotic Fluid Culture for Genetic Testing
88235-Tissue culture for amniotic fluid (if appropriate)
88240-Cryopreservation (if appropriate)

Fibroblast Culture for Genetic Testing
88233-Tissue culture, skin or solid tissue biopsy (if appropriate)
88240-Cryopreservation (if appropriate)

Reflex Tests:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
FBC	Fibroblast Culture for Genetic Test		Profile	No	Yes
AFC	Amniotic Fluid Culture/Genetic Test		Profile	No	Yes
FUSEQ	DNA Sequence, Follow-up Analysis		Profile	No	No

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
FUPCR	Follow Up PCR		Profile	No	No
MCC	Maternal Cell Contamination, B		Profile	No	Yes

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	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		
AFC	16179	Specimen	Alphanumeric		
AFC	16338	Specimen ID	Alphanumeric		
AFC	16793	Source	Alphanumeric		
AFC	16180	Order Date	Alphanumeric		
AFC	CG132	Reason For Referral	Alphanumeric		
AFC	16184	Method	Alphanumeric		
AFC	16187	Interpretation	Alphanumeric		
AFC	16188	Amendment	Alphanumeric		
AFC	16189	Consultant	Alphanumeric		
AFC	16190	Report Date	Alphanumeric		
FUSEQ	17231	Specimen	Alphanumeric		
FUSEQ	17232	Specimen ID	Alphanumeric		
FUSEQ	17233	Source	Alphanumeric		
FUSEQ	17234	Order Date	Alphanumeric		

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
FUSEQ	17235	Result	Alphanumeric		
FUSEQ	17236	Amendment	Alphanumeric		
FUSEQ	17237	Reviewd By	Alphanumeric		
FUSEQ	17238	Released Date	Alphanumeric		
FUPCR	17239	Specimen	Alphanumeric		
FUPCR	17240	Specimen ID	Alphanumeric		
FUPCR	17241	Source	Alphanumeric		
FUPCR	17242	Order Date	Alphanumeric		
FUPCR	17243	Result	Alphanumeric		
FUPCR	17244	Amendment	Alphanumeric		
FUPCR	17245	Reviewed By:	Alphanumeric		
FUPCR	17246	Released Date	Alphanumeric		
MCC	23332	Specimen	Alphanumeric		
MCC	23333	Specimen ID	Alphanumeric		
MCC	23334	Source	Alphanumeric		
MCC	23335	Order Date	Alphanumeric		
MCC	23338	Result	Alphanumeric		
MCC	23337	Amendment	Alphanumeric		
MCC	23341	Reviewed By:	Alphanumeric		
MCC	23339	Release Date	Alphanumeric		

Reference Values:

An interpretive report will be provided.