

Krabbe Disease Second-Tier Newborn Screen, Blood Spot

Test ID: KD2T

Explanation:

On the effective date, the KD2T assay will be obsoleted. The primary recommended alternative testing is the **PSY** (Psychosine, Blood Spot). **KRABZ** (Krabbe Disease, Full Gene Analysis and Large (30 kb) Deletion, Varies) is the recommended follow up for abnormal PSY results.

Primary Recommended Alternative Test:

Psychosine, Blood Spot

Test ID: PSY

Methodology:

Liquid Chromatography Tandem Mass Spectrometry (LC-MS/MS)

Reference Values:

Normal <2 nmol/L

Specimen Requirements:

- Supplies:** Card-Blood Spot Collection (Filter Paper) T493
- Preferred:** Card-Blood Spot Collection (Filter Paper)
- Acceptable:** PerkinElmer 226 filter paper, Munktell filter paper, Whatman protein Saver 903 paper, local newborn screening card, or blood collected in tubes containing EDTA (preferred) or heparin and dried on filter paper
- Specimen Volume:** 2 Blood spots
- Collection Instructions:**
1. Completely fill at least 2 circles on the filter paper card (approximately 100 microliters blood per circle).
 2. Let blood dry on filter paper at ambient temperature in a horizontal position for a minimum of 3 hours.
 3. Do not expose specimen to heat or direct sunlight.
 4. Do not stack wet specimens.
 5. Keep specimen dry.
- Minimum Volume:** 1 Blood spot

Specimen Stability Information:

Specimen Type	Temperature	Time	Special Container
Whole blood	Ambient (preferred)	96 days	FILTER PAPER
	Frozen	96 days	FILTER PAPER
	Refrigerated	96 days	FILTER PAPER

CPT Code:

82542

Day(s) Performed:

Monday through Sunday

Report Available:

1 to 2 days

Follow Up Recommended Alternative Test:**Krabbe Disease, Full Gene Analysis and Large (30 kb)
Deletion, Varies****Test ID:** KRABZ**Methodology:**

Polymerase Chain Reaction (PCR) followed by DNA Sequencing

Reference Values:

An interpretive report will be provided.

Shipping Instructions:

Specimen preferred to arrive within 96 hours of collection.

Specimen Requirements:

Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. Call 800-533-1710 for instructions for testing patients who have received a bone marrow transplant

Submit only 1 of the following specimens:**Specimen Type:** Whole blood**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

Minimum Volume: 1 mL

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

Additional Information: A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

Specimen Type: Skin biopsy

Supplies: Fibroblast Biopsy Transport Media (T115)

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.

Specimen Volume: 4-mm punch

Specimen Stability Information: Refrigerated (preferred)/Ambient

Additional Information: A separate culture charge will be assessed under CULFB / Fibroblast Culture for Biochemical or Molecular Testing. An additional 3 to 4 weeks is required to culture fibroblasts before genetic testing can occur.

Specimen Type: Blood spot

Supplies: Card - Blood Spot Collection (Filter Paper) (T493)

Container/Tube:

Preferred: Collection card (Whatman Protein Saver 903 Paper)

Acceptable: PerkinElmer 226 (formerly Ahlstrom 226) filter paper or blood spot collection card

Specimen Volume: 2 to 5 Blood spots

Collection Instructions:

1. An alternative blood collection option for a patient older than 1 year is a fingerstick. For detailed instructions, see [How to Collect Dried Blood Spot Samples](#).
2. Let blood dry on the filter paper at ambient temperature in a horizontal position for a minimum of 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

Additional Information:

1. Due to lower concentration of DNA yielded from blood spot, it is possible that additional specimen may be required to complete testing.

Minimum Volume: 2 Blood spots

CPT Code:

81406 GALC (galactosylceramidase) (eg, Krabbe disease), full gene sequence

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

88240-Cryopreservation (if appropriate)

Day(s) Performed:

Varies

Report Available:

14 to 20 days

Questions

Contact Melissa Tricker-Klar, Laboratory Resource Coordinator at 800-533-1710.