Newborn Screen Follow-up for Infantile Krabbe Disease

**Decreased GALC and abnormal 2nd-tier test**

**Positive 2nd-tier test Elevated PSY and/or homozygous 30kb deletion**
- Early infantile Krabbe disease
- Immediate referral to Genetics Specialist and HSCT Center for confirmatory testing and evaluation

**Inconclusive 2nd-tier test Normal PSY Heterozygous 30 kb deletion**
- CBGC / Galactocerebrosidase, Leukocytes
- PSY / Psychosine, Blood Spot
- KRABZ / Krabbe Disease, Full Gene Analysis and Large (30 kb) Deletion, PCR*

**GALC deficient PSY elevated Any genotype**
- Referral to Genetics Specialist
- Longterm monitoring for Krabbe disease**

**GALC deficient PSY normal Genotype consistent with later onset Krabbe disease or variant of uncertain significance**
- Not Krabbe

**GALC normal PSY normal Genotype not consistent with Krabbe disease (includes carrier status and GALC pseudodeficiency)**

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GALC=galactocerebrosidase  
PSY=psychosine  
HSCT=hematopoietic stem cell transplant

* GALC deletion/duplication testing should be considered if KRABZ is not informative
** Includes, but may not be limited to, neurologic exam, magnetic resonance imaging (MRI), brain with diffusion tensor imaging (DTI), brainstem auditory evoked responses (BAER), visual evoked potential (VEP), electroencephalography (EEG), nerve conduction, neurocognitive testing, lumbar puncture for cerebrospinal fluid (CSF) protein, psychosine monitoring (blood spot, CSF)