Newborn Screen Follow-up for Krabbe Disease:
Galactocerebrosidase

Newborn screening result:
deprecated galactocerebrosidase (GALC)

Perform the following:
- GALCW / Galactocerebrosidase, Leukocytes
- PSY / Psychosine, Blood Spot OR PSYWB / Psychosine, Blood
- KRABZ / Krabbe Disease, Full Gene Analysis and Large (30kb) Deletion, PCR, Varies
- Consider GALC deletion/ duplication analysis if sequencing not informative

- GALC deficient
- Psychosine (PSY) elevated
- Genotype consistent with Krabbe disease

- Early infantile Krabbe disease
- Immediate referral to Genetics Specialist and hematopoietic stem cell transplant center

- GALC deficient
- PSY intermediate/normal
- Genotype suggestive of Krabbe disease

- Referral to Genetics Specialist
- Lifelong monitoring for Krabbe disease*

- GALC deficient
- PSY abnormal/normal
- Genotype of unknown significance

- Referral to Genetics Specialist
- Consider Saposin A deficiency as a differential diagnosis (PSAP1 full gene sequencing to confirm)

- GALC normal
- PSY abnormal/normal
- Genotype not consistent with Krabbe disease

- Not Krabbe disease
- Consider Saposin A deficiency as a differential diagnosis (PSAP1 full gene sequencing to confirm)

* Neurologic exam, magnetic resonance imaging (MRI) brain with diffusion tensor imaging (DTI), brainstem auditory evoked response (BAEP), visual evoked potential (VEP), electroencephalogram (EEG), nerve conduction, neuro-cognitive testing, lumbar puncture for spinal fluid protein, psychosine monitoring (blood, spinal fluid)