Newborn Screen Follow-up for Krabbe Disease: Galactocerebrosidase

Newborn screening result: decreased 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

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

  Lifelong monitoring for Krabbe disease*
  - 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)

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

* 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)