

## CNMC Newborn Screening Protocols Amid COVID-19 Outbreak

Last Updated: SV 3/23/2020

\*This is a fluid document due to the constant, evolving outbreak situation. Updates and additions can be found on the living [google document](#).\*

**Consider Hold:** *In well babies*, the following borderline/abnormal analytes will be considered non-urgent and recommendation will be to await follow-up visit for labs or intervention until a return to pre-outbreak protocols:

- Any enzyme screen (GAA, IDUA, GLA, GALT or Biotinidase) with history of one normal non-transfused screen for that enzyme (even if normal was collected < 24 hours)
- Any GLA enzyme
- Repeatedly low GALT enzyme with initial normal galactose and subsequent elevation(s) in galactose
- GAA or IDUA (DC or VA) with sequencing detecting  $\leq 1$  pathogenic/likely pathogenic/VUS variants
- Tyrosine (DC or VA) with normal Succinylacetone (SUAC)
- Repeat screen, non-critical Leucines with history of one normal valid screen in well baby
- C5OH (no abnormal biotinidase) or C4 screens
- Abnormal C8, C6, C10 and C8/C10 +/- molecular confirmation of MCADD (DC), these children are all but confirmed and should be treated presumptively via fasting avoidance. CNMC provider will provide ED letter to the family upon request.
- C14+C16 screens (VA) with no C14:1 elevation
- Biotinidase screens: Start 5 mg over-the-counter biotin.
  - Pediatricians can consider Biotinidase enzyme assay if desired but this can wait.

### Plan for Hold Referrals:

- CNMC available to review by phone with pediatrician and/or family.
  - Pediatricians should reach out directly to NBS Genetics Contact (Call 202-476-5000 and ask for Genetics Newborn Screening pager). Telemedicine visits available but these will be non-lab follow-ups.
- CNMC Genetics will reevaluate in 6-8 weeks whether follow-up available or appropriate at that time.

**Pediatrician/Local Action Recommended:** *In well babies*, we will recommend pediatrician-directed follow-up and labs via pediatrician office or lab closer to family's home. Pediatrician can contact CNMC NBS Genetics Contact (Call 202-476-5000 and ask for Genetics Newborn Screening pager) for details but should expect something along the lines of the following:

- 1st-tier GAA or IDUA screens (Maryland-only): We can send pediatrician office Lantern dried blood spot kit and filled out requisition. The office should collect and send the blood spot directly to Perkin Elmer in pre-addressed envelope provided.
  - Email Dr. Christina Grant [cgrant@childrensnational.org] and Dr. Tam Roshan-Lal [troshanlal@childrensnational.org] to arrange.
- Low GALT screen with elevated total galactose: Switch to soy-based formula and obtain Quantitative GALT enzyme (Local Codes for Labcorp: 827640 or Quest 47728X)
  - If GALT Enzyme  $\leq 2.4$ : immediate referral to CNMC via on-call service, (Call 202-476-5000 and ask for Genetics On-Call).
  - If GALT enzyme  $> 2.4$ /interpretation is non-classic galactosemia: Follow-up in 8 weeks via NBS Genetics Contact (Call 202-476-5000 and ask for Genetics Newborn Screening pager) to assess whether follow-up available or appropriate at that time.
- Methionine: obtain Liver Function Panel, Plasma Amino Acids and Plasma Homocysteine
  - If Homocysteine and methionine normal: no follow-up.
  - If Homocysteine normal and methionine elevated: follow-up in 8 weeks via NBS Genetics Contact (Call 202-476-5000 and ask for Genetics Newborn Screening pager) to assess whether follow-up available or appropriate at that time.
  - If Homocysteine elevated: immediate referral to CNMC via on-call service (Call 202-476-5000 and ask for Genetics On-Call).
- Tyrosine (Maryland, no Suac available): obtain Liver Function Panel, Plasma Amino Acids, Mayo SUAC blood spot (SUAC)
  - If liver function and tyrosine normal: no follow-up
  - If liver function normal and tyrosine elevated: follow-up in 8 weeks via CNMC NBS Genetics Contact (Call 202-476-5000 and ask for Genetics Newborn Screening pager) to assess whether follow-up available or appropriate at that time.
  - If liver function (particularly Alk Phos) markedly abnormal and tyrosine elevated: immediate referral/possible direct admission to CNMC via on-call service (Call 202-476-5000 and ask for Genetics On-Call).
- Phenylalanine with Phe/Tyr  $\leq 5$ : obtain Plasma Amino Acids
  - If plasma Phe  $< 360$  umol/L: follow-up in 6-8 weeks via CNMC NBS Genetics Contact (Call 202-476-5000 and ask for Genetics Newborn Screening pager) to assess whether follow-up available or appropriate at that time.
  - If plasma Phe 360-600 umol/L: contact Erin Macleod, 202-476-5179 to discuss possible next steps.

- If plasma Phe > 600: immediately contact Erin Macleod, 202-476-5179 to discuss referral.
- C3
  - Normal C3/C2 (especially with history of ABO incompatibility or jaundice): careful pediatrician evaluation
    - If child is or becomes ill-appearing: immediate referral/possible direct admission to CNMC via on-call service (Call 202-476-5000 and ask for Genetics On-Call).
    - If child well: follow-up in 8 weeks via CNMC NBS Genetics Contact (Call 202-476-5000 and ask for Genetics Newborn Screening pager) to assess whether follow-up available or appropriate at that time.
  - Abnormal C3/C2: careful pediatrician evaluation, obtain Basic Metabolic Panel, Urine Organic Acids and Plasma Acylcarnitines if possible
    - If child is or becomes ill-appearing and/or ammonia or BMP abnormal: immediate referral/possible direct admission to CNMC via on-call service, (Call 202-476-5000 and ask for Genetics On-Call).
    - If child well, labs normal: no follow-up needed.
    - If child well, acylcarnitines show elevated C3: follow-up in 8 weeks via CNMC NBS Genetics Contact (Call 202-476-5000 and ask for Genetics Newborn Screening pager) to assess whether follow-up available or appropriate at that time.
- Low C0: obtain free/total carnitine on baby (and if possible Mom)
  - If baby's total carnitine < 10 umol, contact NBS Genetics Contact (Call 202-476-5000 and ask for Genetics Newborn Screening pager) for direction on starting Levocarnitine supplementation.
  - If baby's total carnitine is below reference range: follow-up in 8 weeks via NBS Genetics Contact (Call 202-476-5000 and ask for Genetics Newborn Screening pager) to assess whether follow-up available or appropriate at that time.
  - If baby's total carnitine normal: no referral necessary.