Transition to Adult Health Care ACT Sheet

Transition is an ongoing process that does not end with transfer of care. The goal of transition of adolescents with chronic medical conditions is to provide uninterrupted, comprehensive, culturally sensitive, coordinated, and developmentally appropriate healthcare. The transition team includes at least the patient and family, and the pediatric, adult PCP, and specialty care providers. ***For the general principles of transition, refer to the 2011 AAP/AAFP/ACP transition clinical report, which includes the recommendation that transition planning begin no later than age 12 and includes a patient readiness assessment.***

N-Acetylglutamate Synthase (NAGS) Deficiency

[Urea Cycle Disorder]

Condition Description: The urea cycle is the enzyme cycle whereby ammonia is converted to urea. NAGS deficiency is an autosomal recessive genetic disorder, in which N-acetylglutamate synthase is defective, interrupting the urea cycle and resulting in hyperammonemia. Hyperammonemia can be very toxic to the brain. Neurocognitive function is variable. Management usually includes a diet low in protein and/or drug therapy.

Clinical Considerations: In affected individuals, excessive protein intake in relation to their diet and/or catabolism due to stress (prolonged fasting, infection, fever, pregnancy and postpartum, surgery, systemic steroids) can lead to hyperammonemia. Valproic acid should be avoided. Immediate clinical evaluation is required when the patient exhibits fever or signs and symptoms of hyperammonemia, such as sleepiness, recurrent vomiting, neurological and psychological findings. The patient should have an acute illness protocol that should be taken to the emergency room (see acute illness protocol). Pregnancies should be considered high risk. There are generally no special considerations with puberty, sexual function and fertility.

THE TRANSITION TEAM SHOULD TAKE THE FOLLOWING ACTIONS:

- **Initiate a dialogue among transition team members and establish an adult medical home.**
- **Facilitate consistency and coordination of care among multiple health care providers as the patient transitions to independent living (to include college, relocation, employment).**
- **Consult with specialists (ideally the metabolic specialist and dietitian caring for the patient) to establish a co-management plan, including input from the patient/family. This care plan should include:**
  - **Nutritional assessment (diet low in protein)**
  - **Drug therapy as indicated (Carbaglu® [carglumic acid])**
  - **Up to date immunizations**
  - **Avoidance of valproic acid and systemic steroids**
- **Confirm the diagnosis by review of the medical record and previous laboratory studies.**
- **Order laboratory studies as indicated (blood ammonia and plasma amino acids).**
- **Identify the patient's health care coverage (including insurance) and access to care.**
- **Assess and address the patient's psychological, behavioral, and social service needs.**
- **Offer health education and genetic counseling concerning future reproductive decisions.**
- **Make patient aware of urea cycle disorders support group.**

Additional Information:

AAP/AAFP/ACP Transition Clinical Report
Transition Toolkit **(New England Consortium of Metabolic Programs)**
Got Transition
NAGS Deficiency
National Urea Cycle Disorders Foundation

Referral (local, state, regional and national):

Clinical Services
Find Genetic Services

LOCAL RESOURCES: Insert State program web site links

State Resource site *(insert program information)*

Name	
URL	
Comments	

APPENDIX: Resources with Full URL Addresses

Acute Illness Protocol
https://www.newenglandconsortium.org/acute-illness

Additional Information:

AAP/AAFP/ACP Transition Clinical Report
https://pediatrics.aappublications.org/content/pediatrics/early/2011/06/23/peds.2011-0969.full.pdf

New England Consortium of Metabolic Programs Transition Toolkit
https://www.newenglandconsortium.org/printable-transition-toolkits

Got Transition
http://www.gottransition.org

Medline Plus
https://medlineplus.gov/genetics/condition/n-acetylglutamate-synthase-deficiency

National Urea Cycle Disorders Foundation
http://www.nucdf.org

Referral *(local, state, regional and national)**:***

Clinical Services
http://www.ncbi.nlm.nih.gov/sites/GeneTests/clinic?db=GeneTests

Find Genetic Services
https://clinics.acmg.net/