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.***

Homocystinuria

(CBS Deficiency)

Condition Description: Methionine from ingested protein is normally converted to homocysteine. Classical homocystinuria is an autosomal recessive genetic disorder due to a deficiency of the vitamin B_6-dependent enzyme cystathionine β-synthase (CBS). In this disorder, homocysteine cannot be metabolized to cystathionine, resulting in elevated levels of homocysteine and methionine.

Clinical Considerations: The clinical complications in untreated or late treated patients can include intellectual disability, ectopia lentis, thromboembolic phenomena, psychiatric symptoms, and osteoporosis. Many patients transitioning to adult care will have been identified and treated with diet and/or betaine from early life, and may have few or none of these complications. In adult years, the major threat is thromboembolic events, and betaine with methionine/protein restriction remains the treatment of choice. Major psychiatric symptoms, including psychosis and severe depression, may be associated with this condition. Osteoporosis is a major concern in adult years. Those patients who are vitamin B_6-responsive will be protected by pharmacologic doses of pyridoxine (vitamin B_6). Pregnancy should be considered high risk because thromboembolic complications, particularly in the post-partum period, represent a major threat and need to be addressed with the consideration of anticoagulation therapy. Estrogen containing oral contraceptives should be avoided due to hypercoagulability. There are generally no other 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 specific for homocystinuria
    - Drug therapy as indicated (betaine [Cystadane®])
    - Up-to-date immunizations
- Confirm the diagnosis by review of the medical record and previous laboratory studies. Document whether the patient is vitamin B_6-responsive.
- Order laboratory studies as indicated (plasma amino acids and total homocysteine).
- 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 homocystinuria support group.

Additional Information:

AAP/AAFP/ACP Transition Clinical Report
Transition Toolkit **(New England Consortium of Metabolic Programs)**
Got Transition

Referral (local, state, regional and national):

Clinical Services
Find Genetic Services

ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

LOCAL RESOURCES: Insert State program web site links

State Resource site *(insert program information)*

Name	
URL	
Comments	

APPENDIX: Resources with Full URL Addresses

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

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/

ACMG
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