

## 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.*

### Galactosemia (GALT Deficiency, Classical Galactosemia)

**Condition Description:** Galactosemia is an autosomal recessive genetic disorder in which a deficiency of galactose-1-phosphate uridylyltransferase (GALT) results in accumulation of galactose-1-phosphate and galactose as well as a secondary metabolite, galactitol, when lactose (milk and milk products) is ingested.

**Clinical Considerations:** Despite neonatal detection and early treatment, adults with galactosemia very frequently have long-term complications that include cognitive deficits, expressive speech difficulty, tremors, depression, anxiety and decreased bone density. Premature ovarian insufficiency (POI) usually occurs within the first decade of life, resulting in amenorrhea, lack of secondary sexual characteristics, and infertility. Lactose-restricted diet for life is recommended, but the value of lactose restriction in adulthood is currently being questioned.

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#### **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 a metabolic specialist and dietitian to establish a co-management plan, including input from the patient/family. This care plan should include:
    - Continuation of a non-dairy diet
    - Consultation with an endocrinologist regarding premature ovarian failure, ideally by age 12
  - Confirm the diagnosis by review of the medical record and previous laboratory studies.
  - Order laboratory studies in consultation with specialists (such as RBC galactose-1-phosphate, urine galactitol, bone density, and for females, gonadotropins and estradiol)
  - Identify the patient's health care coverage (including insurance) and access to care.
  - Assess and address the patient's speech, psychological, behavioral, and social service needs.
  - Offer health education and genetic counseling concerning future reproductive decisions.
  - Make patient aware of galactosemia support group.
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#### **Additional Information:**

[AAP/AAFP/ACP Transition Clinical Report](#)  
[Transition Toolkit](#) (New England Consortium of Metabolic Programs)  
[Got Transition](#)  
[Galactosemia Foundation](#)

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

[Clinical Services](#)  
[Find Genetic Services](#)

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

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LOCAL RESOURCES: Insert State program web site links

State Resource site (insert program information)

Name	<input type="text"/>
URL	<input type="text"/>
Comments	<input type="text"/>

APPENDIX: Resources with Full URL Addresses

*Additional Information:*

AAP/AAFP/ACP Transition Clinical Report

<http://aappolicy.aappublications.org/cgi/reprint/pediatrics;128/1/182.pdf>

New England Consortium of Metabolic Programs Transition Toolkit

<http://newenglandconsortium.org/for-families/transition-toolkit/>

Got Transition

<http://www.gottransition.org>

Galactosemia Foundation

<http://galactosemia.org/>

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

Clinical Services

<http://www.ncbi.nlm.nih.gov/sites/GeneTests/clinic?db=GeneTests>

Find Genetic Services

<http://www.acmg.net/GIS/Disclaimer.aspx>

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