



## Orphan Drugs: Improving the Health of Every Patient

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## American College of Medical Genetics (ACMG)

Key roles in the incorporation of genetics/genomics into the practice of medicine and in conveying accurate information about genetics to patients and the public

More than 1400 MD and PhD clinical and laboratory geneticists and other genetic health care professionals

National Coordinating Center for Regional Genetic Collaboratives funded by the Health Resources and Services Administration (HRSA)

Five-year contract from the National Institute of Child Health and Human Development (NICHD) to establish Newborn Screening Translational Research Network



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Almost all genetic diseases identified by routine newborn screening are rare.

Present significant management challenges for primary healthcare providers.

Contribute to the frustrating and expensive “diagnostic odyssey” for patient and family

ACT (Action) Sheets developed to inform and guide appropriate medical management decisions



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# ACMG ACT Sheet and Algorithm

**Newborn Screening ACT Sheet**  
**(Increased total galactose with normal GALT)**  
**Primary or Secondary Hypogalactosemia**

**Indicated: TOTAL GALACTOSE WITH NORMAL GALT**

**Qualify description:** Children with elevated total galactose (TGal) on newborn screening (NBS) with normal galactose-1-epimerase (GALT) activity on NBS. This finding is associated with primary or secondary hypogalactosemia.

**Prevalence:** Unknown. Estimated to be 1 in 100,000 to 1 in 200,000.

**Genetic heterogeneity:** This finding is associated with primary or secondary hypogalactosemia. The genetic heterogeneity is unknown.

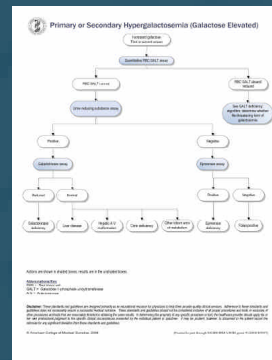
**Prognosis of hypogalactosemia:** The degree of hypogalactosemia will determine the prognosis. Children with primary hypogalactosemia will require lifelong medical management. Children with secondary hypogalactosemia will require medical management until the underlying cause is identified.

**Check for associated conditions:** This finding is associated with primary or secondary hypogalactosemia. Check for associated conditions such as galactosemia, galactose-1-epimerase deficiency, and galactose-4-epimerase deficiency.

**Check for associated symptoms:** This finding is associated with primary or secondary hypogalactosemia. Check for associated symptoms such as vomiting, diarrhea, and failure to thrive.

**Check for associated laboratory findings:** This finding is associated with primary or secondary hypogalactosemia. Check for associated laboratory findings such as elevated TGal, normal GALT, and normal galactose-1-epimerase activity.

**Check for associated clinical findings:** This finding is associated with primary or secondary hypogalactosemia. Check for associated clinical findings such as vomiting, diarrhea, and failure to thrive.



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## **American College of Medical Genetics (ACMG)**

Orphan drugs are essential to proper care of many of the patients with genetic disorders.

Newborn Screening Translation Research Network will provide:

- Collaboration among clinical centers and laboratories
- Development of comprehensive registries of patients
- Implementation of pilot programs and clinical trials
- Establishment of physical or virtual repositories for dried blood spot cards and other medical specimens
- Configuration of information technology (IT) system to aggregate and analyze data



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