Outline for Primary Care Guide for a Positive Newborn Screen

*Please refer to the* [***ACMG ACT Sheets***](https://www.acmg.net/ACMG/Medical-Genetics-Practice-Resources/ACT_Sheets_and_Algorithms.aspx) *as one of your primary research sources.*

**Condition Name**

***Immediate steps upon receiving a positive newborn screen result for a patient***

Abstract

*Required: Provide an abstract of about 125 words. Define the problem or topic addressed in the article. Explain its significance in the broader context of medicine, healthcare, or public health. Describe how the topic is analyzed or reviewed and key insights.*

Key Words

*List keywords for this article. Include other names and acronyms.*

Disorder Category

*As designated by the American College of Medical Genetics and Genomics (ACMG):*

* *Amino Acidemias, Endocrine Disorders, Fatty Acid Oxidation Disorders, Galactosemias, Genetic Disorders, Hemoglobin Disorders, Immunodeficiency Disorders, Lysosomal Storage Diseases, Musculoskeletal Disease, Organic Acidemias*

Screening

Abnormal Finding

*What is the screening finding (analyte) that suggests this diagnosis?*

Tested By

*What test is used in screening for this disorder?*

Description

*Briefly, what is the biological basis of this disorder? General information that a PCP needs to know.*

Clinical Characteristics  
*What this might look like right after birth.*

**With treatment**

*Mix in basic treatment info.*

**Without treatment,**

Incidence

*How common is it?*

Inheritance   
*How is the disorder inherited? Include brief information about family testing if available.*

Primary Care Management

Next Steps After a Positive Screen

* *Use bullets.*
* *If there are common situations resulting in falsely positive newborn screening results, how would I know if that applies here?*
* *What are my first steps, now that I have been notified of the positive screen?*
* *Where can I find help to respond to this positive screen?*
* *Where can I find an expert in the evaluation and management of this condition?*

Confirming the Diagnosis

* *To confirm the diagnosis, work with* [*Newborn Screening Services*](https://www.medicalhomeportal.org/services/category/491)*.*
* *Use bullets.*
* *What diagnostic tests should be ordered, and how should this testing should be coordinated with specialists?*

If the Diagnosis is Confirmed

*How should I help the child and the family? What specialists, therapies, help, or services should I refer the family and patient to? What should I expect from a subspecialty physician or clinic?*

* *Use bullets*
* *For evaluation and ongoing collaborative management, consult*[*Pediatric Genetics*](https://www.medicalhomeportal.org/services/category/337)*.*
* *Refer family to Genetic Testing and Counseling?*

Patient Education

*Required: Include at least one online or downloadable factsheet, brochures, etc., that would be useful for the primary care clinician to share with patients/families.*

Acknowledgments

*optional*

Affiliations

*mandatory*

Declaration of Conflicting Interests

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References

IMPORTANT: *Citations and references follow the* [AMA Manual of Style, 11th Edition.](https://academic.oup.com/amamanualofstyle/book/27941/chapter/207563234?searchresult=1)

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