

# TABLE OF CONTENTS

INTRODUCTION

MEDICAL PROGRAM FOLLOW-UP AND CONSULTANTS

SCREENING TESTS

## AMINO ACID DISORDERS (PKU, MSUD, HCY, TYR I, ASA & CIT)

Introduction

Amino Acid Disorders Screened in Kansas

Clinical Features

Laboratory Screening Tests

Confirmatory Testing

Treatment

Screening Practice Considerations

Medical Consultant

Overview of Follow-up Procedure

Amino Acid Disorder ACT Sheets

Amino Acid Disorder Algorithms

Low Risk Physician's Letter Example

Moderate Risk Physician's Letter Example

High Risk Physician's Letter Example

Physician's Reporting Form Examples

Information for Healthcare Providers

Parent Letter Example

Information for Parents

## BIOTINIDASE DEFICIENCY (BIOT)

Introduction

Clinical Features

Causes of Biotinidase Deficiency

Laboratory Screening Tests

Confirmatory Testing

Treatment

Screening Practice Considerations

Medical Consultant

Overview of Follow-up Procedure

ACT Sheet

Algorithm

Presumptive Physician's Letter Example

Physician's Reporting Form Example

Information for Healthcare Providers

Parent Letter Example

Information for Parents

## CONGENITAL ADRENAL HYPERPLASIA (CAH)

- Introduction
- Clinical Features
- Causes of Congenital Adrenal Hyperplasia
- Laboratory Screening Tests
- Confirmatory Testing
- Treatment
- Medical Consultants
- Overview of Follow-Up Procedure
- ACT Sheet
- Algorithm
- Borderline Physician's Letter Example
- Presumptive Physician's Letter Example
- Physician's Reporting Form Example
- Information for Healthcare Providers
- Parent Letter Example
- Information for Parents

## CONGENITAL HYPOTHYROIDISM (CH or HYPOTH)

- Introduction
- Clinical Features
- Causes of Congenital Hypothyroidism
- Laboratory Screening Tests
- Confirmatory Testing
- Treatment
- Screening Practice Considerations
- Medical Consultants
- Overview of Follow-Up Procedure
- ACT Sheet
- Algorithm
- Borderline Physician's Letter Example
- Presumptive Physician's Letter Example
- Physician's Reporting Form Example
- Information for Healthcare Providers
- Parent Letter Example
- Information for Parents

## CYSTIC FIBROSIS (CF)

- Introduction
- Clinical Features
- Causes of Cystic Fibrosis
- Laboratory Screening Tests
- Confirmatory Testing
- Treatment
- Screening Practice Considerations
- Medical Consultants
- Overview of Follow-Up Procedure
- ACT Sheet
- Algorithm
- Initial Positive Physician's Letter Example
- Repeat Positive Physician's Letter Example
- Physician's Reporting Form Example
- Information for Healthcare Providers
- Parent Letter Example
- Information for Parents

## FATTY ACID OXIDATION DISORDERS (MCADD, VLCAD, LCHADD, TFP & CUD)

- Introduction
- Fatty Acid Oxidation Disorders Screened in Kansas
- Clinical Features
- Laboratory Screening Tests
- Confirmatory Testing
- Treatment
- Screening Practice Considerations
- Medical Consultant
- Overview of Follow-up Procedure
- Fatty Acid Oxidation Disorder ACT Sheets
- Fatty Acid Oxidation Disorder Algorithms
- Low Risk Physician's Letter Example
- Moderate Risk Physician's Letter Example
- High Risk Physician's Letter Example
- Physician's Reporting Form Examples
- Information for Healthcare Providers
- Parent Letter Example
- Information for Parents

## GALACTOSEMIA (GALT)

- Introduction
- Clinical Features
- Laboratory Screening Tests
- Confirmatory Testing
- Treatment
- Screening Practice Considerations
- Medical Consultants
- Overview of Follow-Up Procedure
- ACT Sheet
- Algorithm
- Borderline Physician's Letter Example
- Presumptive Physician's Letter Example
- Physician's Reporting Form Example
- Information for Healthcare Providers
- Parent Letter Example
- Information for Parents

## HEMOGLOBINOPATHIES

- Introduction
- Clinical Features
- Laboratory Tests
- Confirmatory Testing
- Treatment
- Carrier Detection
- Screening Practice Considerations
- Infant Transfusion
- Medical Consultants
- Follow-Up Procedure
- Inheritance Patterns
- ACT Sheets
- Algorithm
- Physician's Letter Example - Hgb Traits
- Physician's Letter Example - Hgb Disease
- Physician's Reporting Form Example
- Information for Healthcare Providers
- Parent Letter Example - Hgb Traits
- Parent Letter Example - Hgb Disease
- Information for Parents

## ORGANIC ACID DISORDERS (IVA, GA-1, HMG, MCD, MUT, Cbl A&B, 3MCC, PROP & BKT)

- Introduction
- Clinical Features
- Causes of Organic Acid Disorders
- Laboratory Screening Tests
- Confirmatory Testing
- Treatment
- Screening Practice Considerations
- Medical Consultants
- Overview of Follow-up Procedure
- ACT Sheets
- Algorithms
- Low Risk Physician's Letter Example
- Moderate Risk Physician's Letter Example
- High Risk Physician's Letter Example
- Physician's Reporting Forms
- Information for Healthcare Providers
- Parent Letter Example
- Information for Parents

## PHENYLKETONURIA (PKU)

- Introduction
- Clinical Features
- Laboratory Tests
- Confirmatory Testing
- Treatment
- Screening Practice Considerations
- Variant Forms of PKU
- Maternal PKU and Hyperphenylalaninemia
- Medical Consultants
- Overview of Follow-Up Procedure
- Follow-Up Flow Chart
- ACT Sheet
- Algorithm
- Low Risk Physician's Letter
- Moderate Risk Physician's Letter
- High Risk Physician's Letter
- Physician's Reporting Form
- Information for Healthcare Providers
- Parent Letter
- Information for Parents

## SPECIAL HEALTH SERVICES

- Services Available - General Information
- Eligibility
- Services for Newborn Screening Disorders

## SCREENING PRACTICES

- Screening Kit Information
- Completion of the Form
- Sample Screening Kit
- Timing of Testing
- Obtaining the Specimen
- Mailing Instructions
- Reporting of Results
- Reporting and Follow-up of "Presumptive" Results
- Problems in Screening Practices
  - Testing Before Discharge
  - Protein and Lactose Ingestion Prior to Testing
  - Testing Before Transfer of Infant to Another Unit
  - Completion of the Form
  - Transfusions
  - Infants Who Are Never Tested
  - Specimen Inadequacy
  - Parent Refusal of Testing

## APPENDICES

- A Kansas Law Pertaining to Newborn Screening - House Bill No. 2255
- B Kansas Newborn Screening Regulations
- C Parent Refusal Form
- D Screening the Critically Ill and Premature Infant
- E Kansas Newborn Screening Brochure
- F Kansas Newborn Screening Collection Form
- G Newborn Screening Specimen Collection Technique
- H Unsatisfactory Specimen Codes