



SOUTH CAROLINA
DEPARTMENT OF
PUBLIC HEALTH

REQUEST FOR DIAGNOSIS

Attention healthcare facilities and physicians: South Carolina Laws 44-37-30 and 44-37-35 and Regulation 60-80 mandate the reporting of inborn metabolic errors and hemoglobinopathies to the SC Department of Public Health.

Note: Federal HIPAA legislation allows disclosure of protected health information, without consent of the individual, to public health authorities for the purpose of preventing or controlling disease. (HIPAA 45 CFR §164.512).

Email completed form to nbsfollowup@dph.sc.gov or Fax to 803-898-0337

Patient's Name:

DOB:

Mother's Name:

PCP Name/Practice:

Referred to (Specialist Name/Practice):

Status:

☐ Cleared

☐ Diagnosed

☐ Expired

Diagnosis/ICD-10:

Relevant Test Results:

Medication/Treatment:

Additional Comments:

Name of Person Completing Form:

Email Address:

Contact or Office Phone Number:

Today's Date:

List of Newborn Screening Conditions tested on the South Carolina Bloodspot Screening Panel

2,4 Dienoyl-CoA Reductase Deficiency (DE RED)	Holocarboxylase Synthetase Deficiency (MCD)
2-Methyl-3-Hydroxybutyric Acidemia (2M3HBA)	Homocystinuria (HCY)
2-Methylbutyrylglycinuria (2MBG)	Hypermethioninemia (MET)
3-Hydroxy-3-Methylglutaric Aciduria (HMG)	Isovaleric Acidemia (IVA)
3-Methylcrotonyl-CoA Carboxylase Deficiency (3-MCC)	Krabbe Disease (globoid cell leukodystrophy)
3-Methylglutaconic Aciduria (3MGA)	Long-Chain L-3 Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)
Argininemia (ARG)	Malonic Acidemia (MAL)
Argininosuccinic Aciduria (ASA)	Maple Syrup Urine Disease (MSUD)
Benign Hyperphenylalaninemia (H-PHE)	Medium/Short-Chain L-3 Hydroxyacyl-CoA Dehydrogenase Deficiency (M/SCHAD)
Beta-Ketothiolase Deficiency (BKT)	Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
Biopterin Defect in Cofactor Biosynthesis (BIOPT-BS)	Medium-Chain Ketoacyl-CoA Thiolase Deficiency (MCAT)
Biopterin Defect in Cofactor Regeneration (BIOPT-REG)	Methylmalonic Acidemia (Cobalamin Disorders) (Cbl A,B)
Biotinidase Deficiency (BIOT)	Methylmalonic Acidemia (Methylmalonyl-CoA Mutase Deficiency) (MUT)
Carnitine Acylcarnitine Translocase Deficiency (CACT)	Methylmalonic Acidemia with Homocystinuria (Cbl C, D, F)
Carnitine Palmitoyltransferase Type I Deficiency (CPT-IA)	Mucopolysaccharidosis Type-I (MPS I)
Carnitine Palmitoyltransferase Type II Deficiency (CPT-II)	Mucopolysaccharidosis Type-II (MPS II)
Carnitine Uptake Defect (CUD)	Primary Congenital Hypothyroidism (CH)
Citrullinemia, Type I (CIT)	Propionic Acidemia (PROP)
Citrullinemia, Type II (CIT II)	S, Beta-Thalassemia (Hb S/βTh)
Classic Galactosemia (GALT)	S, C Disease (Hb S/C)
Classic Phenylketonuria (PKU)	Severe Combined Immunodeficiency (SCID)
Congenital Adrenal Hyperplasia (CAH)	Sickle Cell Anemia (Hb SS)
Cystic Fibrosis (CF)	Spinal Muscular Atrophy (SMA)
Fabry Disease (FD)	T-cell related lymphocyte deficiencies
Galactoepimerase Deficiency (GALE)	Trifunctional Protein Deficiency (TFP)
Galactokinase Deficiency (GALK)	Tyrosinemia, Type I (TYR I)
Glutaric Acidemia, Type I (GA-1)	Tyrosinemia, Type II (TYR II)
Glutaric Acidemia, Type II (GA-2)	Tyrosinemia, Type III (TYR III)
Glycogen Storage Disease Type II (Pompe)	Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)
Hemoglobinopathies (Var Hb)	X-Linked Adrenoleukodystrophy (ALD)

NEWBORN SCREENING REQUEST FOR DIAGNOSIS

Instructions for Completing 4474-ENG-DPH

Purpose: The form is to be used for submitting newborn screening diagnosed case information to the Department of Public Health Newborn Screening Program for patient case follow-up documentation.

Audience: Completed by healthcare providers after receiving an infant's abnormal newborn screening results.

Instructions:

1. Enter the patient's name and date of birth (DOB).
2. Enter Mother's first and last name.
3. Enter the patient's primary care physician's name and practice.
4. If the patient was referred to a specialist, enter the specialist's name and practice.
5. Mark the appropriate status of the patient:
 - a. **Cleared:** Secondary testing and patient assessment is normal. The patient **does not** have a newborn screening condition identified by dried blood spot screening.
 - b. **Diagnosed:** The patient was diagnosed with a condition that was identified by newborn screening dried blood spot testing.
 - c. **Expired:** Infant mortality.
6. Enter the patient's final diagnosis/ICD-10 code as indicated. A comprehensive list of newborn screening disorders on the South Carolina newborn screening bloodspot panel is located on the second page.
7. Enter or attach any relevant test results.
8. Enter the name(s) of any related treatment or medication(s) the patient was prescribed.
9. Enter any additional comments that are relevant.
10. Enter the full name and contact information of the person filling out the form.
11. Enter a contact email address in the event there are additional questions.
12. Enter the office phone number or contact phone number of the person filling out the form.
13. Enter the date the form is being filled out.
14. Email or fax the completed form to nbsfollowup@dph.sc.gov or 803-898-0337 and keep a copy for your records.

Office Mechanics and Filing: Form will be retained following DPH records retention schedule 18810, "Newborn Screening Follow-up Documentation Records," Records Group Number: 169. The form will be retained in the Public Health Laboratory (PHL) Newborn Screening Follow-Up Office for 9 years. After this time period is met, the information will be stored at the State Records Center for an additional 10 years, and then destroyed.