DEPARTMENT OF PUBLIC HEALTH AND ENVIRONMENT

Laboratory Services Division

NEWBORN SCREENING AND SECOND NEWBORN SCREENING

5 CCR 1005-4

[Editor's Notes follow the text of the rules at the end of this CCR Document.]

NEWBORN SCREENING REGULATIONS

1.1 Under the authority contained in Sections 25-4-801 through 25-4-804 and 25-4-1001 through 25-4-1006 (not including Section 25-4-1004.7) C.R.S. (1998), the following rules and regulations are established.

1.2 Definitions: The following terms, whenever used in or referred to in these regulations, shall have the following respective meanings, unless a different meaning clearly appears from the context:

"Department" shall mean the Colorado Department of Public Health and Environment.

"Laboratory" shall mean the Colorado Department of Public Health and Environment Laboratory.

"Initial Newborn Screening Specimen" shall mean specimen collected from a newborn prior to discharge, but in all cases within 48 hours after birth for the purpose of conducting screening tests.

1.3 Procedures

1.3.1 Births in Institutions: The blood specimens of newborns born in institutions and all other specimens taken in conformity with the law and these regulations will be sent to the Laboratory for testing. Follow up specimens from newborns with positive screening tests will be obtained and tested as necessary for proper diagnosis.

1.3.1.1 The hospital or institution or the chief medical staff officer or other person in charge thereof will cause an initial newborn screening specimen to be obtained from every newborn born therein as late as possible before discharge, but no later than 48 hours of age.

1.3.1.2 The specimen shall consist of capillary blood collected by heel puncture or alternate method authorized by the Laboratory, directly upon special blotter paper furnished by the Laboratory. All circles shall be saturated with blood from one side of the blotter only. The specimen submitter will provide, on the attached form, all information requested by the Laboratory. The specimens, after air drying, will be forwarded to the Laboratory within 24 hours of collection, by courier or overnight delivery if available.

1.3.1.3 If the newborn is to receive a blood transfusion, then the specimen for newborn screening is to be obtained prior to this procedure .

1.3.2 Births Outside Institutions: The physician, nurse midwife, or other health professional attending a birth outside a hospital, shall be responsible for the collections and forwarding of the specimen described in 1.3.1.2 above. In the absence of a health professional, any

other person attending the birth, or in the absence of any person so attending, the father or mother, or in the absence of the father and the inability of the mother, the person in charge of the premises where the birth occurred shall be responsible.

1.4 Testing and Reporting: The prescribed tests will be initiated by the Laboratory within 24 hours of receipt of the specimen, weekends and holidays excepted. The Laboratory shall report as follows:

1.4.1 Reports of normal test results will be sent to the submitting agency within seven working days.

1.4.2 Abnormal test results will be reported immediately by telephone to the physician of record and to designated consultants. In case of inability to identify or locate a physician of record, the abnormal test result will be reported to the hospital or submitting agency which originated the specimen, or, if the birth did not occur in a health facility, to the father or mother.

1.4.3 Unsatisfactory specimens or specimens with equivocal results will be reported immediately to the submitting agency which originated the specimen with an explanation of the results. The submitting agency responsible for the newborn's care at the time of the report will cause another specimen to be forwarded at the appropriate time.

1.4.4 The submitting agency that originated the specimen shall forward the Newborn Screening results to the health care provider responsible for the newborn's care within the time frame of 1.4.1 and 1.4.3 above.

1.5 Quality Control and Education

1.5.1 The Laboratory shall have available for review a written quality assurance program plan covering all aspects of laboratory activity.

1.5.2 The Laboratory shall make available educational materials and training concerning specimen collection to all submitting agencies.

1.6 List of Conditions for Newborn Screening

1.6.1 The Laboratory shall conduct screening tests for the following conditions:

1.6.1.1 Phenylketonuria

1.6.1.2 Congenital Hypothyroidism

1.6.1.3 Hemoglobinopathies

1.6.1.4 Galactosemia

1.6.1.5 Cystic Fibrosis

1.6.1.6 Biotinidase Deficiency

1.6.1.7 Congenital Adrenal Hyperplasia

1.6.1.8 Medium Chain Acyl-CoA dehydrogenase deficiency

1.6.1.9 Very Long Chain Acyl-CoA dehydrogenase deficiency

1.6.1.10 Long-Chain L-3-Hydroxy Acyl-CoA dehydrogenase deficiency

1.6.1.11 Trifunctional protein deficiency

1.6.1.12 Carnitine Acyl-carnitine translocase deficiency

1.6.1.13 Short Chain Acyl-CoA dehydrogenase deficiency

1.6.1.14 Carnitine palmitoyltransferase II deficiency

1.6.1.15 Glutaric acidemia Type 2

1.6.1.16 Arginosuccinic acidemia

1.6.1.17 Citrullinemia

1.6.1.18 Tyrosinemia

1.6.1.19 Hypermethionemia

1.6.1.20 Maple Syrup urine disease

1.6.1.21 Homocystinuria

1.6.1.22 Isovaleric acidemia

1.6.1.23 Glutaric acidemia Type 1

1.6.1.24 3-hydroxy-3-methylglutaryl-CoA Lyase deficiency

1.6.1.25 Multiple Carboxylase deficiency

1.6.1.26 3-methylcrotonyl-CoA carboxylase deficiency

1.6.1.27 3-methylglutaconic aciduria

1.6.1.28 Methylmalonic acidemias

1.6.1.29 Propionic acidemia

1.6.1.30 beta-Ketothiolase deficiency

1.6.1.31 Carnitine uptake defect

1.6.1.32 Arginase deficiency

1.6.1.33 Malonic acidemia

1.6.1.34 Carnitine palmitoyltransferase deficiency 1A

1.6.1.35 Severe Combined Immunodeficiency

RULES AND REGULATIONS OF THE EXECUTIVE DIRECTOR COLORADO DEPARTMENT OF PUBLIC HEALTH AND ENVIRONMENT

IMPLEMENTATION OF SECOND NEWBORN SCREENING

1.1 Under the authority contained in Section 25-4-1004.5(3) C.R.S., the following Rules and Regulations are established.

1.2 Definitions

"Department" shall mean the Colorado Department of Public Health and Environment.

"Executive Director" shall mean the executive director of the Colorado Department of Public Health and Environment.

"Laboratory" shall mean the Colorado Department of Public Health and Environment Laboratory.

"Initial newborn screening specimen" shall mean specimen collected from a newborn prior to discharge, but in all cases within ~~seven days~~ 48 HOURS after birth for the purpose of conducting screening tests.

"Second newborn screening specimen" shall mean a specimen collected from a newborn between eight and 14 days after birth, but in no case less than 72 hours or greater than 30 days after birth, for the purpose of conducting screening tests.

1.3 Procedures

1.3.1 The parent(s) or other legal guardian(s) of the newborn shall be advised of the necessity of the second newborn screening test.

1.3.1.1 Births in Institutions: It shall be the responsibility of the hospital or institution or the chief medical staff officer or other person in charge thereof to advise, verbally and in writing, such as by written information made available from the Department, the parent(s) or other legal guardian(s) of the newborn that it is necessary to have a second newborn screening test performed.

1.3.1.2 Births outside Institutions: It shall be the responsibility of the physician, nurse midwife, lay midwife, or other health professional attending a birth outside a hospital to advise, verbally and in writing, such as by written information made available from the Department, the parent(s) or other legal guardian(s) of the newborn, of the necessity of the second newborn screening.

1.3.2 The attending health care provider shall collect or require the specimen be collected from all newborns at the first post partum appointment, but in no case less than 72 hours or greater than 30 days after birth. The specimen shall consist of capillary blood collected by heel puncture or alternate method authorized by the Laboratory, directly upon special blotter paper furnished by the Laboratory. All circles shall be saturated with blood from one side of the blotter only. The ~~specimen collector~~ SUBMITTER will provide, on the ~~provided~~ ATTACHED form, all information requested by the Laboratory. The specimens, after air drying, shall be forwarded to the Laboratory within 24 hours of collection by first class mail, COURIER, or ~~other appropriate means~~ OVERNIGHT DELIVERY.

1.4 Testing and Reporting: The prescribed tests will be initiated by the Laboratory within 24 hours of receipt of the specimen, weekends and holidays excepted. The Laboratory shall report as follows:

1.4.1 Reports of normal test results will be sent to the submitting agency within seven working days.

1.4.2 Abnormal test results will be reported immediately by telephone to the physician of record and to designated consultants. In case of inability to identify or locate a physician of record, the abnormal test result will be reported to the submitting agency which originated the specimen, or, if the birth did not occur in a health facility, to the father or mother.

1.4.3 Unsatisfactory specimens or specimens with equivocal results will be reported immediately to the submitting agency which originated the specimen with an explanation of the results. The health care provider responsible for the newborn's care at the time of the report will cause another specimen to be forwarded at the appropriate time.

1.4.4 The submitting agency that originated the specimen shall forward the newborn screening results to the health care provider responsible for the newborn's care.

1.5 Quality Control and Education

1.5.1 The Laboratory shall have available for review a written quality assurance program plan covering all aspects of testing and reporting second specimens.

1.5.2 The Laboratory shall make available educational materials and training concerning specimen collection to submitting agencies.

1.6 List of Conditions for Second Newborn Screening

1.6.1 The Laboratory shall conduct screening tests for the following conditions:

1.6.1.1 Phenylketonuria

1.6.1.2 Congenital Hypothyroidism

1.6.1.3 Hemoglobinopathies

1.6.1.4 Galactosemia [1]

1.6.1.5 Cystic Fibrosis [1]

1.6.1.6 Biotinidase Deficiency [1]

1.6.1.7 Congenital Adrenal Hyperplasia

1 These disorders need not be tested again unless:

a) an unsatisfactory specimen was submitted for first screen testing, or

b) an abnormal result was obtained on first screen testing, or

c) no record of a satisfactory first screen specimen submission can be ascertained.

Editor's Notes

History

Newborn Screening Regulation 1.6 eff. 01/30/2008.

Second Newborn Screening Regulation 1.6 eff. 04/01/2008.

Entire Rule eff. 04/30/2011.