Regulatory Analysis Form	This space for use by IRRC RECEIVED 2001 APR 18 PM 2:01 REVIEW COMMISSION IRRC Number: 2188
(1) Agency Department of Health	
(2) I.D. Number (Governor's Office Use) Reg. No. 10-137	
(3) Short Title Metabolic Diseases of the Newborn	
(4) PA Code Cite 28 Pa. Code ch. 27 28 Pa. Code ch. 28 28 Pa. Code ch. 501	(5) Agency Contacts & Telephone Numbers Primary Contact: Jack W. Means, Jr. 717-783-8143 Secondary Contact: Maryann McCarthy, RN 717-783-8143
(6) Type of Rulemaking (Check One) X Proposed Rulemaking __ Final Order Adopting Regulation __ Final Order, Proposed Rulemaking Omitted	(7) Is a 120-Day Emergency Certification Attached? X No _ Yes: By the Attorney General _ Yes: By the Governor

(8) Briefly explain the regulation in clear and non-technical language.

The Department of Health (Department), with the approval of the State Advisory Health Board (Board), is proposing to amend the regulations currently set out at 28 Pa. Code ch. 28, to incorporate changes required as a result of amendments to the Act of September 9, 1965 (P.L. 497, No. 251), known as the Newborn Child Testing Act (35 P.S. §§621-625) and to add two additional screens as permitted by 35 P.S. §623(b). The proposed regulations amend current regulations by adding maple syrup urine disease (MSUD), hemoglobin diseases, galactosemia and congenital adrenal hyperplasia (CAH) to the list of diseases at 28 Pa. Code §28.2 (relating to newborn diseases listed) for which newborn children are routinely screened. The proposed regulations also update existing procedures for screening and follow-up testing for those diseases which are currently listed, as well as those which would be added by these proposed regulations. Finally, the proposed regulations clarify the circumstances under which a health care provider would be responsible for collecting the initial and any repeat blood filter paper specimens for testing.

The Department is also proposing minor amendments to its regulations at Chapter 27 (relating to reporting laboratory results indicative of certain diseases) and Chapter 501 (relating to newborn infant care policies and procedures in birth centers) of Title 28 of the Pennsylvania Code. These supplemental amendments are necessary in order to ensure conformity with the requirements of the expanded Newborn Screening Program.

(9) State the statutory authority for the regulation and any relevant state or federal court decisions.

The Department obtains its authority to promulgate these regulations from several sources. Generally, the Disease Prevention and Control Law of 1955 (35 P.S. §521.1 et seq.) provides the Advisory Health Board with the authority to issue rules and regulations on a variety of issues relating to communicable and non-communicable diseases, including the methods of reporting diseases, the contents of those reports and the health authorities to whom diseases are to be reported. (35 P.S. §521.16(a)). §16(b) of the Disease Prevention and Control Law of 1955 (35 P.S. §521.16(b) gives the Secretary of Health (Secretary) the authority to review existing regulations and make recommendations to the Board for changes the Secretary considers to be desirable.

The Department also finds general authority for the promulgation of its regulations in the Administrative Code of 1929 (71 P.S. §51 et seq.). Section 2102(g) of the Administrative Code (71 P.S. 532(g)) gives the Department the general authority. Section 2111(b) of the Administrative Code (71 P.S. §541(b)) provides the Advisory Health Board with additional authority to promulgate regulations deemed by the Board to be necessary for the prevention of disease, and for the protection of the lives and the health of the people of the Commonwealth. That section further provides that the regulations of the Board shall become the regulations of the Department.

The Department's specific authority for promulgating the regulations relating to newborn screening and follow-up is found in the Newborn Child Testing Act (Act) (35 P.S. §§621-625). Section 5 of the Act (35 P.S. §625) provides the Department, with the approval of the Board, with the authority to promulgate regulations for the implementation and administration of the Act. Section 3(b) of the Act (35 P.S. §623) provides the Department, with the approval of the Board, with the authority to establish by regulation those diseases for which newborn children shall be tested and the methods for testing and disseminating test results. Section 4(b) of the Act (35 P.S. §624) provides the Department with the authority to establish by regulation the methods of procurement of blood specimens of newborn children by health care providers.

(10) Is the regulation mandated by any federal or state law or court order, or federal regulation? If yes, cite the specific law, case or regulation, and any deadlines for action.

The language in the regulations concerning the addition of MSUD and hemoglobin diseases is based upon the requirements of the Newborn Child Testing Act, 35 P.S. §§ 621-625. Galactosemia and CAH are added pursuant to 35 P.S. §§ 623(b) which permits the addition to the list by regulation of any other disease approved for such inclusion by the Department and the Board. Certain procedures, including the time frames in which to collect the specimens, are based upon the requirements for detecting MSUD, CAH and galactosemia and preventing harm to the newborn child.

(11) Explain the compelling public interest that justifies the regulation. What is the problem it addresses?

Expansion of the Commonwealth's Newborn Screening Program to include hemoglobin diseases, MSUD, galactosemia and CAH and the clarification of uniform procedures governing screening, is intended to facilitate early detection and treatment of diseases and conditions in the newborn child, and will result in earlier referral for diagnosis and treatment of newborn children identified with presumptive abnormal tests for these diseases. Early detection of, treatment for, these diseases will save the lives of many newborn children, and will also significantly reduce long term health care costs for the families of affected children, as well as for public and private medical providers and insurers.

Sickle cell disease is the most common of the hemoglobin diseases and is most often found among individuals of African American origin (affecting one in every 400 black infants). Approximately 72 newborns with sickle cell disease and 28 newborns with other forms of hemoglobin diseases will be diagnosed in Pennsylvania each year. There is a high incidence of serious bacterial infections and mortality in young children with sickle cell disease. A 1986 study organized by the National Institutes of Health demonstrated that early detection of children with sickle cell disease followed by treatment with penicillin will reduce the incidence of pnuemonoccocal sepsis by 89 percent. Early identification of sickle cell disease will save the lives of many infants with disease. For others, early and improved medical care and family education will reduce time spent by the children in the hospital, and time lost from work by parents.

MSUD is a genetic disorder most prevalent in members of the Mennonite sect. One in 225,000 newborn children nationwide is identified with MSUD each year. The incidence of MSUD in Pennsylvania, however, is three times greater than the national average, and it is expected that two children will be identified with MSUD annually in the Commonwealth. MSUD is a very volatile condition that afflicts infants within days of birth, and which, if not diagnosed and treated quickly, will cause irreversible mental and physical damage, or death.

Untreated children die or are virtually incapacitated, requiring lifetime care. With early diagnosis and the institution of treatment that includes a special diet, affected children can survive with normal intelligence. As a result of screening and early treatment, costs for hospitalization of these children are greatly reduced, and long term costs for residential care are virtually eliminated.

Galactosemia is a genetic metabolic condition, which effects the body's ability to utilize certain sugars. The most common forms of galactosemia may result in death from sepsis within the first weeks of life or mental retardation in those who survive. Prompt diagnosis and intervention can prevent further damage. One in 60,000 newborn is identified with classical galactosemia. When other forms of galactosemia are included, such as Duarte and Los Angeles, the rate increases to one newborn in 16,000 identified with a form of galactosemia.

CAH is a genetic disorder that involves a deficiency of enzymes that catalyzes severe salt wasting, hypertension, stunted growth and pseudohermaphroditism. Mortality from "adrenal crisis" is high. Proper medical treatment resets the abnormal balance of hormones and permits near normal development. One in 16,000 newborn is identified with CAH.

The Department initiated pilot screening programs for hemoglobin diseases and MSUD in several parts of the Commonwealth on June 1, 1990, and May 15, 1991, respectively. The pilot projects confirmed that addition of these diseases to the screening program was warranted. These pilot projects also provided a basis for evaluating and refining laboratory procedures and follow-up procedures for the entire screening program. Statewide screening for hemoglobin diseases began on September 28, 1992, and for MSUD on March 31, 1993. The proposed regulations reflect these additions; they also reflect the need to obtain specimens in a shorter time period following the child's birth than the current regulations require.

(12) State the public health, safety, environmental or general welfare risks associated with non-regulation.

The proposed regulation adds two diseases to the current regulations as mandated by the Newborn Child Testing Act, 35 P.S. §§ 621-625, and the others pursuant to 35 P.S. §§ 623(b) which permits the addition to the list by regulations of any other disease approved for such inclusion by the Department and the Board. The proposed regulations also change the time frames for specimen collection currently set out in the regulations. The current time frames included in regulation are too long given the extreme danger of disability and death that MSUD can pose for a newborn child. Early detection and treatment of all the disease and conditions of newborn children listed in the regulations will prevent severe mental and physical damage to newborn children, and will save long term health care costs of providing for children severely impaired by these diseases and conditions.

The proposed regulations also provide authoritative procedures for collecting and testing newborn blood filter paper specimens, and for the follow-up of abnormal test results correctly and in a timely fashion so that disability or death of the newborn child can be averted. Due to the importance of efficient collection and testing of screening specimens for phenylketonuria (PKU), primary congenital hypothyroidism, hemoglobin diseases, MSUD, galactosemia and CAH, the proposed regulations will designate specific responsibility for the collection and follow-up of newborn screening tests by health care providers, and will specify time of collection and mailing which will assist prompt follow-up of newborn children with one of these diseases or conditions.

(13) Describe who will benefit from the regulation. (Quantify the benefits as completely as possible and approximate the number of people who will benefit.)

The approximately 150,000 children born in the Commonwealth yearly and their families will benefit from the proposed regulations because with early detection of PKU, MSUD, hemoglobin diseases, primary congenital hypothyroidism, galactosemia and CAH children will be referred for diagnosis and treatment within days of birth. Early medical intervention can prevent life threatening illness and permanent medical and physical disability. Approximately 90 children are diagnosed with hemoglobin diseases, 2 children with MSUD, 10 children with CAH and 10 with galactosemia in the Commonwealth each year.

The health care providers of the Commonwealth will benefit by having regulations setting out their responsibilities, including appropriate time frames for collection of specimens. Health care providers have repeatedly asked for formal guidance from the Department on this matter.

(14) Describe who will be adversely affected by the regulation. (Quantify the adverse effects as completely as possible and approximate the number of people who will be adversely affected.)

To the extent that health care providers will be required to comply with more stringent time frames for collection of specimens, and follow more precise procedures for the collecting of specimens, these providers may be said to be adversely affected. Again, however, screening for hemoglobin diseases and MSUD began across the Commonwealth in 1992 and 1993 respectively; screening for PKU and hypothyroidism has been occurring since 1965 and 1978 respectively. Screening for galactosemia and CAH are proposed to commence on a voluntary basis in 2000-2001. Given the necessity of early detection of MSUD, common prudence and risk management would have recommended a more stringent screening time frame than that required by existing regulations in any case.

(15) List the persons, groups or entities that will be required to comply with the regulation. (Approximate the number of people who will be required to comply.)

All newborns born in Pennsylvania must be screened for PKU, sickle cell disease (hemoglobinopathies), and MSUD according to the Newborn Child Testing Act, 35 P.S. §§ 621-625. The existing regulations added screening for hypothyroidism as well.

The proposed regulation adds MSUD, hemoglobin diseases, galactosemia and CAH to current regulations, which require screening for PKU and hypothyroidism. The statute does, however, allow parents to exercise a religious exemption to the screening requirements if they so choose. See 35 P.S. § 623(c). All health care providers (defined by the proposed regulation to include those providing maternity and newborn services) throughout Pennsylvania are currently required by statute and regulation to collect blood filter paper specimens from newborn children and to submit them to the Department's contract laboratory for testing. The proposed regulation reflects the 1992 amendments to the law which added sickle cell disease (hemoglobinopathies) and MSUD to the list of diseases and conditions for which newborn children are screened. Health care providers are currently, and will continue to be required to collect and submit a blood filter paper specimen and assist with follow-up of abnormal, inconclusive, and unacceptable test results for these diseases in accordance with the proposed regulation. Laboratories will be required by the proposed regulations to report four additional diseases, hemoglobin diseases, MSUD, galactosemia and CAH, to the Department.

This will impact the Department's contracted laboratory, which performs all the initial testing of newborn screening specimens, and which currently does test and report findings of hemoglobin diseases and MSUD to the Department through its contract with the Department. The contract laboratory will have to expand testing to include galactosemia and CAH, although it presently screens nearly 80% of Pennsylvania's newborns for these conditions through its supplemental program. Other laboratories may be sent specimens for follow-up testing; these requirements would impact those laboratories.

(16) Describe the communications with and input from the public in the development and drafting of the regulation. List the persons and/or groups who were involved, if applicable.

During the initial drafting of the proposed regulation comments were solicited from the following: physicians specializing in the care of infants with metabolic, endocrine or hematologic conditions; laboratory personnel; and the state Newborn Screening Technical Advisory Committee. Strong public demand to obtain newborn screening for additional conditions was made evident through individual efforts, the public media, and service organizations.

(17) Provide a specific estimate of the costs and/or savings to the regulated community associated with compliance, including any legal, accounting or consulting procedures which may be required.

It is probable that since the passage of the Newborn Child Testing Act, 35 P.S. §§ 621-625, in 1992, health care providers have incurred modest costs in personnel and staff time for the provision of follow-up services for repeat specimens or abnormal test results related to MSUD and hemoglobin diseases, when necessary. Specimen collection, testing, and follow-up for MSUD and sickle cell and other abnormal hemoglobins of clinical significance have been fully integrated with existing procedures for PKU and hypothyroidism. Blood specimens for MSUD, hemoglobin diseases, galactosemia and CAH testing will be taken from the same heel stick as the specimens currently collected for hypothyroidism and PKU screening, and be placed on the same filter paper. The number of unacceptable specimens and abnormal results for which follow-up reporting and consultation services are required is expected to be low; the proposed regulation is intended to reduce the number of unacceptable specimens and abnormal results by clarifying and standardizing procedures and timing for specimen collection.

Neither health care providers nor families of the newborn children who are tested are currently charged for laboratory screening or follow-up test costs. It should be noted that the proposed regulation includes requirements which were statutorily mandated in 1992, so that any increased costs associated with the additions of MSUD and hemoglobin diseases should have occurred at that time.

Without early identification, diagnosis, and treatment, newborn children affected by these diseases are at risk for early death, severe mental impairment, or at the least, medical problems. If death does not occur, the family of a child in whom these diseases go undetected may incur high costs related to hospitalization, intensive care, the need for residential care, and special education needs. The cost to institutionalize one child for one year is approximately $80,000. The number of affected newborn children who were not appropriately diagnosed and treated would multiply this cost. In the Commonwealth in 1999, there were 24 children diagnosed with PKU, four with MSUD, 45 with congenital hypothyroidism, and 99 with hemoglobin diseases (72 sickle cell disease and 27 other abnormal hemoglobinopathies). The Department expects approximately 10 newborns per year to be born with galactosemia and CAH based on recent laboratory results. Not every child diagnosed with one of these diseases is necessarily institutionalized; however, based upon $80,000 yearly cost per case, if we assume half of the newborns identified with one of these conditions is institutionalized, the cost could exceed $7 million dollars each year. These figures do not take into account anguish suffered by families whose children die or who are severely mentally impaired.

(18) Provide a specific estimate of the costs and/or savings to local governments associated with compliance, including any legal, accounting or consulting procedures which may be required.

Local governments are not affected by the proposed regulation.

(19) Provide a specific estimate of the costs and/or savings to state government associated with the implementation of the regulation, including and legal, accounting, or consulting procedures which may be required.

Expansion of the Commonwealth's Newborn Screening Program to include MSUD and hemoglobin diseases pursuant to the requirements of the Newborn Child Testing Act, 35 P.S. §§ 621-625, has resulted in some increased program costs to the Department. The Department estimates that the additional cost for screening and follow-up for these two additional diseases is approximately $1,300,000 per year. However, state government will also realize savings through the early detection, timely referral and follow-up treatment of children afflicted with these additional conditions. Newborn screening helps to prevent various forms of developmental disabilities and their associated medical costs that are incurred for services such as in-home supports, institutionalization and other ongoing medical care. Public resources, such as state/federal Medicaid funds and state-only Mental Health/Mental Retardation program funds, often pay these costs.

(20) In the table below, provide an estimate of the fiscal savings and costs associated with implementation and compliance for the regulated community, local government and state government for the current year and five subsequent years.

	2000/ 2001	2001/ 2002	2002/ 2003	2003/ 2004	2004/ 2005	2005/ 2006
SAVINGS:	$ * * *	$	$	$	$	$
Regulated Community	$0	0	0	0	0	0
Local Government	$ N/A	N/A	N/A	N/A	N/A	N/A
State Government	$ 0	0	0	0	0	0
Total Savings	$0	0	0	0	0	0
COSTS:	$					
Regulated Community	$0	0	0	0	0	0
Local Government	$ N/A	N/A	N/A	N/A	N/A	N/A
State Government	$1,300,000	1,300,000	1,300,000	1,300,000	1,300,000	1,300,000
Total Costs	$1,300,000	1,300,000	1,300,000	1,300,000	1,300,000	1,300,000
REVENUE LOSSES:	$ * * *					
Regulated Community	$ 0	0	0	0	0	0
Local Government	$ N/A	N/A	N/A	N/A	N/A	N/A
State Government	$0	0	0	0	0	0
Total Revenue Losses	$0	0	0	0	0	0

(20a) Explain how the cost estimates listed above were derived.

* * * The Commonwealth's Newborn Screening Program implemented statewide screening for hemoglobin diseases and MSUD in 1992 and 1993, respectively, in response to the mandates of the Newborn Child Testing Act, 35 P.S. §§621-625. Health care providers have been complying with these reporting requirements since that time. Any increased costs should have been incurred at the initiation of this screening. In addition, 80% of the providers already screen for galactosemia and CAH through a voluntary private supplemental screening program.

Regulatory Analysis Form				
(20b) Provide the past three year expenditure history for programs affected by the regulation.				
Program	FFY 96	FFY 97	FFY 98	FFY 99
Newborn Screening and Follow-up	$3,157,950	$3,264,480	$3,098,769	$2,571,742

(21) Using the cost-benefit information provided above, explain how the benefits of the regulation outweigh the adverse effects and costs.

Detecting and providing timely referral, follow up treatment on abnormal results through newborn screening helps to prevent various forms of developmental disabilities, including mental retardation and other life-long chronic, debilitating conditions and their associated medical needs. The cost-benefit of providing newborn screening, referral and follow up treatment is significant in that it saves public resources, such as state/federal Medicaid funds and state-only Mental Health/Mental Retardation program funds, due to the avoided costs later in life that are incurred for services such as in-home supports, institutionalization and ongoing and costly medical care.

(22) Describe the nonregulatory alternatives considered and the costs associated with those alternatives. Provide the reasons for their dismissal.

No non-regulatory approaches were considered. There are existing regulations setting out the requirements for screening and follow-up for PKU and hypothyroidism in newborn children set out at 28 Pa. Code ch. 28. These regulations were authorized by the Newborn Child Testing Act, 35 P.S. §§ 621-625; amendments to this act in 1992 mandated expansion of the Commonwealth's Newborn Screening Program to include testing for MSUD, and sickle cell disease (hemoglobinopathies). The Department is amending the existing regulations to include four additional diseases of the newborn, and to clarify screening procedures. Other abnormal hemoglobins of clinical significance, galactosemia and CAH are added pursuant to 35 P.S. §§ 623(b) which permits the addition to the list by regulation of any other disease approved for such inclusion by the Department and the Board.

(23) Describe alternative regulatory schemes considered and the costs associated with those schemes. Provide the reasons for their dismissal.

No other regulatory approaches were considered. There are existing regulations setting out the requirements for screening and follow-up for PKU and hypothyroidism in newborn children set out at 28 Pa. Code ch. 28. These regulations were authorized by the Newborn Child Testing Act, 35 P.S. §§ 621-625; amendments to this act in 1992 mandated expansion of the Commonwealth's Newborn Screening Program to include testing for MSUD and sickle cell hemoglobinopathies. The Department is amending the existing regulations to include four additional diseases of the newborn, and to clarify screening procedures. Other abnormal hemoglobins of clinical significance, galactosemia and CAH are added pursuant to 35 P.S. §§ 623(b) which permits the addition to the list by regulation of any other disease approved for such inclusion by the Department and the Board.

(24) Are there any provisions that are more stringent than federal standards? If yes, identify the specific provisions and the compelling Pennsylvania interest that demands stronger regulation.

There are no federal requirements for the screening of the conditions and diseases of the newborn child.

(25) How does this regulation compare with those of other states? Will the regulation put Pennsylvania at a competitive disadvantage with other states?

This proposed regulation does not bear upon the competitive advantage of the Commonwealth. All states are currently working together to ensure that all newborn children are screened for a minimum number of diseases and conditions. In fact, Pennsylvania screens for very few diseases and conditions compared to other states in the nation.

(26) Will the regulation affect existing or proposed regulations of the promulgating agency or other state agencies? If yes, explain and provide specific citations.

As part of this proposed rulemaking, the Department is proposing minor amendments to 28 Pa. Code Chapters 27 (relating to Communicable and Noncommunicable Diseases) and 501, § 49 (relating to newborn infant care policies and procedures in birth centers). The minor revisions to Chapter 27, "Communicable and Noncommunicable Diseases", include adding MSUD, sickle cell disease, galactosemia and CAH to the list of reportable diseases in § 27.2 (relating to reportable diseases), § 27.22b (relating to reporting of laboratory results indicative of certain infections or conditions), § 27.22d (relating to reporting of laboratory results indicative of certain infections or conditions) and § 27.30 (relating to reporting results of metabolic disease testing in the newborn child.

(27) Will any public hearings or information meetings be scheduled? Please provide the dates, times, and locations, if available.

The Department is not planning any hearings at the present time. The Department has proposed a thirty (30) day comment period after the regulations are published as proposed in the Pennsylvania Bulletin.

(28) Will the regulation change existing reporting, record keeping, or other paperwork requirements? Describe the changes and attach copies of forms or reports which will be required as a result of implementation, if available.

The proposed regulations require health care providers to notify parents or guardians of newborn children when a repeat specimen is required for an abnormal or inconclusive test result or an unacceptable specimen. The proposed regulations also place additional paperwork responsibilities on the testing laboratory and health care providers, because of the addition of four diseases to the list of disease and conditions of newborn children for which testing is required. These requirements are minimal, however. The proposed regulations make laboratories responsible for reporting MSUD, sickle cell disease, galactosemia and CAH to the Department under revisions to the communicable disease regulations at 28 Pa. Code ch. 27. The screening test results for the four new diseases will be reported on the same report form currently being used for PKU and hypothyroidism. Further, the testing laboratory operates under a contract with the Department, and receives additional recompense for additional work. To the extent that other laboratories may do additional confirmatory or follow-up testing, they will be required to comply with these reporting requirements, as they would currently do for PKU and hypothyroidism.

Under the proposed regulation, health care providers will be required to submit data regarding specimen collection to the Department semi-annually; the existing regulations require only that such data be maintained.

Paperwork requirements within the Department itself will remain unchanged. The addition of MSUD, hemoglobin diseases of clinical significance, galactosemia and CAH to the list of diseases tracked by the screening program has increased the number of instances in which follow-up of abnormal results is required; however, follow-up has been occurring since the implementation of the expanded screening program for hemoglobin diseases and MSUD began in 1992 and 1993, respectively.

(29) Please list any special provisions which have been developed to meet the particular needs of affected groups or persons including, but not limited to, minorities, elderly, small businesses, and farmers.

These proposed regulations are intended to protect the health of all newborn children, and, therefore, no special provisions were necessary. The statute upon which the regulations are based already has a provision, which permits parents who may have religious objections to the test to request an exemption on those grounds. See 35 P.S. § 623(c).

(30) What is the anticipated effective date of the regulation; the date by which compliance with the regulation will be required; and the date by which any required permits, licenses or other approvals must be obtained?

The Department is proposing that the regulations become effective upon their final publication in the Pennsylvania Bulletin. Adoption of the proposed regulation is essential to ensure uniform specimen collection, testing, and follow-up procedures by health care providers throughout the Commonwealth. Compliance with the proposed regulation requires no additional required permits, licenses, or approvals.

(31) Provide the schedule for continual review of the regulation.

The efficacy and appropriateness of the proposed regulation will be reviewed through monitoring by Department staff in the Bureau of Family Health and the Bureau of Laboratories. The Bureau of Laboratories will continue to routinely conduct quality assurance reviews of the laboratory performing the screening tests, which is under contract with the Department. Newborn Screening Program staff in the Bureau of Family Health will routinely review reports of test results from the screening laboratory, and reports by hospitals of specimens collected and forwarded for testing. In this way, the Department will be able to determine whether modifications in the screening process are necessary, and can take steps to initiate those modifications.

FACE SHEET
FOR FILING DOCUMENTS
WITH THE LEGISLATIVE REFERENCE BUREAU

(Pursuant to Commonwealth Documents Law)

2188

RECEIVED
2001 APR 18 PM 2:01
REVIEW COMMISSION

DO NOT WRITE IN THIS SPACE

Copy below is hereby approved as to form and legality. Attorney General	Copy below is hereby certified to be a true and correct copy of a document issued, prescribed or promulgated by:	Copy below is hereby approved as to form and legality. Executive or independent Agencies
BY _____ DEPUTY ATTORNEY GENERAL	DEPARTMENT OF HEALTH (AGENCY)	BY _____
APR 04 2001 DATE OF APPROVAL	DOCUMENT/FISCAL NOTE NO. 10-137	3/26/01 DATE OF APPROVAL
	DATE OF ADOPTION: _____	(Deputy General Counsel) (~~Chief Counsel, Independent Agency~~) (Strike inapplicable title)
	BY: Robert S. Zimmerman, Jr. Robert S. Zimmerman, Jr.	
☐ Check if applicable. Copy not approved. Objections attached.	TITLE: Secretary of Health	☐ Check if applicable. No Attorney General approval or objection within 30 days after submission.

PROPOSED RULEMAKING
DEPARTMENT OF HEALTH

TITLE 28. HEALTH AND SAFETY

[28 PA. CODE CHS 27, 28 AND 501]

NEWBORN DISEASE SCREENING AND FOLLOW-UP

Notice is hereby given that the Department of Health (Department), with the approval of the State Advisory Health Board (Board), proposes to amend 28 Pa. Code Chapter 28 (relating to metabolic diseases of the newborn). The Department also proposes to amend specific sections of 28 Pa. Code Chapters 27 (relating to communicable and noncommunicable diseases) and 501 (relating to birth centers), as made necessary by the changes being proposed to Chapter 28. The proposed amendments are set forth in Annex A hereto.

PURPOSE OF THE REGULATIONS

The Department is proposing to amend its regulations to incorporate changes to the Newborn Screening and Follow-Up Program (Program) required as a result of amendments to the Newborn Child Testing Act (Act) (35 P.S. §§ 621 – 625), made by the act of July 9, 1992 (P.L. 398, No. 86). The amendments to the Act add maple syrup urine disease (MSUD) and sickle-cell hemoglobinopathies (disease and trait) to the list of diseases for which routine screening of newborns is conducted, provide for the addition to the list by regulation of any other disease approved for such inclusion by the Department and the Board, and require a screening and follow-up program to identify and treat newborn children with one of the diseases listed in the Act or identified by regulation. 35 P.S. § 623. The Department is given the authority under the Act to promulgate regulations, with the approval of the Board, to carry out these requirements. 35 P.S. § 625. The Department is also proposing to amend the regulations to include additional screens for galactosemia and congenital adrenal hyperplasia (CAH) pursuant to its authority to add to the list of diseases for which routine screening of newborns is to be conducted, by regulation, any other disease approved for such inclusion by the Department and the Board. The Department is further proposing to amend the regulations to screen for hemoglobinopathies (hemoglobin diseases) other than sickle cell hemoglobinopathies because the detection of other hemoglobin diseases, some of which may be life threatening, is unavoidable with the testing methodology currently available.

The Department's Bureau of Family Health, through the Program, began administering a statewide newborn screening program for the detection of phenylketonuria (PKU) in 1965. Screening for hypothyroidism was added to the Program in 1978. Detection and follow-up of newborn children with these two diseases have been conducted pursuant to regulations promulgated by the Department and the Board under authority of the Disease Prevention and Control Law of 1955 (No. 500, P.L. 1510) (35 P.S. §§521.1-521.21) and the Act. These regulations appear at 28 Pa. Code §28.1 et seq.

Amendment of the Act to include sickle cell hemoglobinopathies and MSUD in the list of diseases of the newborn child, for which screening is to be done, requires expansion of the Program. To meet these requirements, the Department implemented statewide screening for sickle cell hemoglobinopathies and MSUD on September 28, 1992, and March 22, 1993,

respectively. Screening for galactosemia and CAH is currently occurring on a voluntary basis.

To reflect the expansion of the Program as required by the Act, the proposed regulations add MSUD and hemoglobin diseases (sickle cell hemoglobinopathies and other clinically significant abnormal hemoglobin) to the list of diseases at 28 Pa. Code § 28.2 (relating to metabolic diseases listed) for which newborns are routinely screened. It is proposed that the title of that section be revised to "Newborn diseases listed." Galactosemia and CAH are not required by the Act, but would be added by the Department with Board approval and through regulation as permitted by the Act. The proposed regulations also update existing procedures for screening and follow-up testing for those diseases which are currently listed, as well as the four added by these proposed regulations. The proposed regulations clarify the circumstances under which a health care provider is responsible for collecting the initial and any repeat blood specimens for testing. Perhaps most importantly, the proposed regulations include new time frames for specimen collection and transmission to the testing laboratory, which reflect the need for speedy results to alleviate the deadly consequences of MSUD, CAH and galactosemia, commensurate with the need for accuracy in testing, which requires collection to be made no earlier than 24 hours after birth.

The Department is also proposing minor amendments to 28 Pa. Code Chapters 27 and 501. Both sets of minor revisions are necessary to ensure that no inconsistencies exist between the updated requirements of the expanded Program and other Department regulations.

Major revisions to Chapter 27 were published as proposed at 30 Pa. B. 2717-2752 (May 27, 2000). The minor revisions to that chapter being proposed in Annex A will be re-evaluated upon final passage of the proposed amendments published on May 27, 2000, and any necessary changes will be incorporated into the final version of the regulations currently being proposed.

REQUIREMENTS OF THE REGULATIONS

CHAPTER 28. METABOLIC DISEASES OF THE NEWBORN

The following is a discussion of the major amendments, additions, and deletions proposed to the regulations governing screening for the diseases of the newborn in Chapter 28. First, the Department is proposing to change the name of this chapter to "Screening and follow-up for diseases of the newborn" to more accurately reflect the nature of the Program. The Act authorizes the Department, with the approval of the Board, to establish screening requirements for diseases leading to mental retardation or physical disabilities, some of which, like hemoglobin disease, are not necessarily metabolic in nature.

Section 28.1. Definitions.

Several terms used in the current regulations are confusing, outdated, or inadequately defined. The Department proposes to revise and replace those terms and definitions with language which reflects the current practice of the Program, and which more adequately reflects the procedures being used. For example, the Department has chosen to replace the term "initial presumptive positive test result," used to denote a test result that indicates the probable presence of disease, with the term "presumptive abnormal test result." The proposed term and its accompanying definition eliminate all reference to a positive or negative result and redesignate screening results as simply normal or abnormal. This change emphasizes that the Program is a screening program, not a diagnostic one. The term "presumptive positive" indicates the confirmation of disease, something that the Program is not intended to do. The term "abnormal" is more appropriate for a screening program as it indicates a screening result, not a diagnosis.

The Department also proposes to eliminate the term "phenylketonuria program clinic" in favor of a term which reflects the expansion of the Program. This term would be replaced by "treatment center." "Treatment center" encompasses clinics seeing referrals for all diseases diagnosed through the Program.

The proposed regulations also introduce other new terms warranting definition. Other terms that would be added to the definition section include: "abnormal confirmatory test result," "abnormal screening test result," "health care provider," "hemoglobin disease," "hospital," "newborn screening program," "presumptive abnormal test result," "repeat specimen," "repeat test," "specimen collection form," and "specimen collection kit." These terms reflect the current operation of the Program.

Additional terms that would be removed from this section are "bureau," "confirmatory test specimen," "health care facility," "hypothyroid referral clinic laboratory," and "initial test."

Section 28.2. Metabolic diseases listed.

The Department is proposing to change the title of this section to "Newborn diseases listed" to remove the inaccurate impression that only the diseases for which newborn children are screened under the Program are metabolic diseases. The diseases for which newborn children are screened under the Program are not all metabolic in nature, therefore, the Department proposes deleting all references in this section and other sections to "metabolic diseases" and replacing it with "newborn diseases."

In this section the Department proposes to add CAH, galactosemia, hemoglobin disease and MSUD to the list of diseases for which mandatory newborn screening is conducted throughout the Commonwealth. It sets out the requirements contained in the Act and reflects screening practices in the Commonwealth following the amendment of the Act in 1992.

The term "hemoglobin disease" encompasses sickle cell hemoglobinopathies (disease and trait) as defined in the Act as well as other hemoglobin diseases and their carrier state (trait). While the primary purpose of neonatal screening of hemoglobin is to identify infants with sickle cell disease, the current testing methodology available for hemoglobin screening also identifies infants with other potentially life threatening abnormal hemoglobin conditions. The Department believes these other hemoglobin diseases should be reported to the health care provider and parent so that, if confirmed, treatment can be initiated.

Sickle cell disease is a disorder that changes the shape of red blood cells as a result of abnormal hemoglobin. This disease, which predominantly affects persons of African-American and Mediterranean descent, may be inherited if both parents carry the sickle cell trait. Early detection of sickle cell disease, followed by prompt medical treatment, will save the lives of many infants with the disease, reduce the high incidence of bacterial infections which would otherwise afflict these children, and decrease the number and length of hospital admissions for these children. Further, detection of sickle cell trait, which is identified during the screening for sickle cell disease, enables the child and his or her family to receive the necessary education and counseling to prevent passage of sickle cell disease to future offspring.

MSUD, the other disease added by the Act in 1992, is a serious genetic disorder most prevalent in members of the Mennonite sect. The incidence of MSUD in Pennsylvania is three times greater than the national average. This disease affects the way the body processes protein, resulting in a blood toxicity, which interferes with brain functions. If not diagnosed and treated within seven days of birth, MSUD can cause severe and irreversible mental retardation and physical incapacitation requiring lifetime care, or death. However, with early diagnosis and treatment affected children can survive. In addition to vastly improving the quality of life and chances for survival for these children, early diagnosis drastically reduces hospital costs and virtually eliminates costs otherwise required for residential care.

Galactosemia is a genetic metabolic condition, which affects the body's ability to break down galactose, a simple sugar found in milk products and many formulas. The most common forms of galactosemia may result in death from sepsis within the first weeks of life or mental retardation in those who survive. Prompt diagnosis and intervention can prevent further damage.

CAH involves a deficiency of enzymes that catalyzes severe salt wasting, hypertension, stunted growth and incorrect sex assignment in female newborns. Mortality from adrenal

crisis is high. Prompt diagnosis and intervention leads to proper medical treatment, which replaces the deficient balance of hormones and permits near normal development.

Section 28.3. Tests to be performed.

This section would be repealed. The text in subsection (a) of this section would be deleted. With technological advancements enabling the detection and treatment of many more diseases and conditions, identifying a host of methodologies in regulation is counterproductive, as some will surely become dated. The Department will prescribe methods and procedures to be used and the standards of accuracy and precision required for each test through the process of selecting a testing laboratory.

The text in subsection (b) would be transferred, without substantive change, to §28.12 (relating to religious objections), as new subsection (a). This reorganization would clarify existing regulations by grouping all provisions concerning religious objections to testing under a single section.

Section 28.4. Standards for collecting and testing specimens.

This section would be repealed. The proposed regulations would incorporate the substantive portion of this section in amendments to proposed §28.21 (relating to responsibility). The title of that section would be changed to "Responsibility for collecting and testing initial and repeat specimens."

Section 28.5. Confidentiality.

This section would be new. As proposed, it sets out confidentiality requirements for health care providers, the testing laboratory, the Department, and any other person or entity involved in the Program. Under the Disease Prevention and Control Law of 1955, under which reporting of these diseases of the newborn is made to the Department, all information obtained pursuant to that law, or used by the Department to take action on the information, is strictly confidential. To make it clear that all information maintained by the Program is governed by confidentiality provisions even if there is some question as to whether the information is governed by the Disease Prevention and Control Law of 1955, the Department has chosen to explicitly state its confidentiality requirements in this regulation. It has always been the Department's policy, under the Disease Prevention and Control Law of 1955, to permit the release of certain aggregate statistical information when no identifying

information is involved, or to release information concerning an individual upon the tendering of the appropriate consent forms. This proposed regulation reflects that policy.

As proposed, the text in subsection (a)(3) follows closely the language in 35 P.S. §10101 (relating to individual consent) which permits a minor who is eighteen years of age or older, or has graduated from high school, or has married, or has been pregnant to give effective consent for medical, dental and health services. A minor who can consent to health care must be able to access his or her records in order to receive that care. This subsection, as proposed, makes that possible.

Section 28.11. Informing the parent or guardian.

This section would remain largely unchanged. The proposed changes to this section would reflect current Program terminology.

Section 28.12. Religious objections.

The text from §28.3(b) would be transferred to subsection (a) of this section. The existing text, without substantive change, would become subsection (b).

Section 28.21. Responsibility.

The Department is proposing to change the name of this section to "Responsibility for collecting and testing initial and repeat test specimens" to clarify that this section refers to the responsibility of health care providers to collect initial and repeat specimens.

The Department also proposes to expand the section itself to clarify the circumstances under which the health care provider would have responsibility for collecting from newborn children the initial and any repeat specimens required. The proposed regulation specifically states that a birth center or hospital would be responsible for collecting all necessary specimens from each newborn delivered at the facility. When a newborn is delivered in a place other than in a birth center or hospital, the health care practitioner who delivered the newborn would be responsible for collecting all necessary specimens.

This section also would be amended to expressly state that it is the responsibility of the health care provider to collect the specimen as set forth in the regulations, to submit properly completed specimen collection forms along with the blood filter paper specimens, and to assist with follow-up of inconclusive, presumptive abnormal, abnormal, and unacceptable test results.

Section 28.22. Timing of initial specimen collection and handling in health care facilities.

The Department is proposing to change the name of this section to "Timing of initial specimen collection by birth centers or hospitals" to accurately reflect the purpose of this section.

The regulation as it is currently written requires that health care facilities (which includes only birth centers and hospitals in the current regulations) collect the initial blood filter paper specimen from the newborn between the fifth and sixth day of age. The birth center or hospital may wait until the newborn is as old as nine days to collect the initial blood filter paper specimen if the newborn's medical condition is unstable. The birth center or hospital may also collect the initial specimen earlier if circumstances, such as early discharge of the newborn from the facility, warrant.

In some cases, these time frames could hamper effective screening. Screening results are less reliable if blood specimens are collected before 24 hours of age because substances in the blood which are measured for detection of these diseases fluctuate significantly within the first 24 hours after birth. If the newborn is discharged before 24 hours of age, screening results may be inaccurate. Repeat testing is necessary in such cases to ensure accuracy of the screening result and efficacy of the Program.

Further, the requirement for screening for MSUD, CAH and galactosemia requires revisions to the time frames currently set out in regulation. Infants with MSUD must be identified by the seventh to fourteenth day of life to initiate effective treatment. Failure to determine whether a newborn has MSUD by the seventh day after birth could result in the death of that newborn child. Similarly, an infant born with CAH may go into adrenal crisis and die within days of birth or an infant born with galactosemia may die from sepsis within the first weeks of life. The current time frames must be altered to reflect this urgency. Given the absolute necessity of early detection in order to initiate treatment for all newborn diseases for which the Department screens, the Department proposes to revise the time frames in this section to require collection of the initial specimen at as close to 48 hours of age as possible but not later than 72 hours.

Finally, the Department proposes to move §28.23(b) (relating to timing of initial specimen by health care practitioners) to subsection (b) of this section. This subsection addresses the hospital's responsibility to obtain a specimen from a child who was delivered somewhere other than a hospital, who is later admitted into a hospital, and who has no record of results of an approved screening test for the newborn diseases listed in §28.2 (relating to metabolic diseases listed). This subsection, therefore, more properly belongs in the section of the regulations which sets out the responsibilities of birth centers and hospitals, not in §28.23 which sets out responsibilities of health care practitioners.

Section 28.23. Timing of initial specimen collection and handling for home births.

The Department is proposing to change the name of this section to "Timing of initial specimen collection by health care practitioners" to accurately reflect the purpose of this section.

This section would be amended so that it would no longer use the term "home birth" to define the circumstance under which the health care practitioner has responsibility for specimen collection. The responsibility for specimen collection by the health care practitioner would be triggered by a birth "other than in a hospital or birth center." The proposed amendments to §28.22 (relating to timing of initial specimen collection by birth centers or hospitals) and this section would mirror the dichotomy proposed in §28.21 (relating to responsibility for collecting and testing initial and repeat specimens); births would be separated into birth center or hospital births and non-birth center or hospital births, with birth centers and hospitals having responsibility for specimen collection for birth center or hospital births and practitioners having responsibility for specimen collection for non-birth center or hospital births. Regardless of which health care provider has the responsibility for collection of the initial specimen, however, that specimen is to be collected from the newborn as close to 48 hours of age as possible but not later than 72 hours of age.

Section 28.24. Negative test results.

The Department is proposing to change the name of this section to "Normal test results" to accurately reflect current Program terminology.

As proposed, all references to "negative test results" would be deleted and replaced with "normal test results." These changes would be made to reflect the changes in Program terminology to "normal" or "abnormal" test results rather than "positive" or "negative" test results.

Section 28.25. Followup recall specimens.

The Department is proposing to change the name of this section to "Circumstances requiring repeat specimens" to accurately reflect the purpose of this section.

As it is presently written, this section requires health care facilities (birth centers and hospitals) and practitioners to collect a "recall" specimen when the initial specimen is unsuitable for testing or the test results are inconclusive. The Department is proposing to retain the requirements outlined above, but to change the term to "repeat" specimen.

Proposed subsection (b) would also add a circumstance in which a repeat specimen is required; a birth center or hospital that discharges a newborn prior to 24 hours of age would be required to collect a repeat specimen. Repeat testing is necessary in this situation because, while it is necessary to obtain a specimen before early discharge in case the infant is not available for testing following discharge, the specimen obtained at less than 24 hours of age will not produce a definitively valid result. Testing should be repeated automatically by the health care provider without consulting with the Department. Finally, this section, as proposed, includes a new subsection (c), which would give the Department discretion to require a repeat specimen when the results of the initial screening are abnormal.

Section 28.26. Timing of recall specimen collection, handling and reporting.

The Department is proposing to change the name of this section to "Timing of repeat specimen collection" to accurately reflect the purpose of this section.

As proposed, subsection (a) would require a birth center or hospital to collect a repeat specimen from a newborn child who was discharged before 24 hours of age as close to 48 hours of age as possible, but not later than 72 hours of age. As proposed, subsection (b) would require that a health care provider collect a repeat specimen within 72 hours of receipt of notice from the Department or testing laboratory that the initial specimen was unacceptable or yielded an inconclusive result.

The Department is proposing that the text of subsection (c) be deleted as it is no longer relevant due to the changes being proposed in subsections (a) and (b), and that the text of subsection (d) be transferred, without substantial change, to subsection (c).

Section 28.27. Followup of presumptive positive test results.

The Department is proposing to change the name of this section to "Abnormal screening test results" to accurately reflect Program terminology and the purpose of this section.

The Department has determined that this section as it is currently written provides inadequate direction for effective response to the full range of abnormal results. The existing section requires follow-up by the health care provider only for results which are presumptively positive, that is, results at the highest level of abnormality. The narrow scope of this provision has created ambiguity with regard to the responsibility of the health care provider for follow-up of only slightly abnormal test results.

The Department proposes, therefore, to amend this section to specify general procedures for the health care provider to follow upon receipt of notification from the Department that the

results of the screening test were abnormal. Under this section, as revised, the Department would determine the appropriate level of follow-up based upon the nature of the abnormal result and instruct the health care provider accordingly. Further, the regulation, as amended, would assign responsibility directly to the health care provider for promptly notifying a parent or guardian of the newborn child with an abnormal test result and arranging for follow-up services. Finally, the revisions to this section would place primary responsibility on the health care provider for locating a parent or guardian of the child.

As amended, this section will also be revised to consolidate and allow for deletion of the text in §28.30 (relating to phenylketonuria) and §28.31 (relating to hypothyroidism). In all cases, the Department will assist the health care provider with and make available confirmatory testing. If the result of the confirmatory test is abnormal, the Department will assist with referral for diagnosis, treatment and other follow-up services for the newborn child through designated treatment centers or clinical specialists.

Section 28.29. Confirmatory test specimen required.

This section would be repealed. Section 28.25 (relating to circumstances requiring repeat specimens) would now address when repeat specimens are required.

Section 28.30. Phenylketonuria.

This section would be repealed. The subject matter now addressed in this section would be addressed in §28.27 (relating to abnormal screening test results).

Section 28.31. Hypothyroidism.

This section would be repealed. The subject matter now addressed in this section would be addressed in §28.27 (relating to abnormal screening test results).

Section 28.41. Recordkeeping requirements.

Amendments to this section would add language to reflect the current practice of health care providers of collecting and forwarding data semiannually to the Department, including the aggregate number of specimens collected and the reasons that certain specimens are not collected.

CHAPTER 27. COMMUNICABLE AND NONCOMMUNICABLE DISEASES

The proposed minor revisions to Chapter 27 include adding CAH, galactosemia, MSUD, PKU, primary congenital hypothyroidism and sickle cell disease in children under 5 years of age as reportable diseases in §27.2 (relating to reportable diseases). The Department also proposes adding CAH, galactosemia, MSUD and sickle cell disease in children under 5 years of age as diseases to be reported, and changing hypothyroidism to primary congenital hypothyroidism and phenylketonuria to PKU in children under 5 years of age in §27.22(b) (relating to reporting laboratory results indicative of certain infections and conditions) and revising what reports need to be submitted to the Department's Division of Maternal and Child Health under §27.22(d) (relating to reporting laboratory results indicative of certain infections and conditions). As proposed, these amendments would not require the reporting under these sections of any hemoglobin disease other than sickle cell disease, as it is the most severe. The identification of other hemoglobin diseases through screening will assist physicians with diagnosis and eliminate confusion with iron deficiency anemia, however, there would be no benefit to having them reported under these sections.

Further, the Department proposes revising §27.30 (relating to reporting results of metabolic disease testing to the newborn child) to reflect the change to the name of Chapter 28.

CHAPTER 501. BIRTH CENTERS

The proposed revisions to §§501.3 and 501.49 (relating to reports/contact person; and newborn infant care policies and procedures) of Chapter 501, "Birth Centers," bring these sections into conformity with the proposed procedures and time frames for specimen collection in the proposed §§28.21 (relating to responsibility for collecting and testing initial and repeat specimens), 28.22 (relating to timing of initial specimen collection by birth centers or hospitals), 28.25 (relating to circumstances requiring repeat specimens), 28.26 (relating to the timing of repeat specimen collection) 28.27 (relating to abnormal screening test results), 28.28 (relating to followup of symptoms consistent with newborn diseases), and 28.41 (relating to recordkeeping requirements).

AFFECTED PERSONS

The proposed regulations will affect all health care providers providing care to pregnant women and newborn children in Pennsylvania, as well as the treatment centers and any laboratory with which the Department contracts to provide the screening services. Health care providers will be required to collect blood filter paper specimens in accordance with updated procedures, assist the Department with follow-up of certain test results, and forward

data on specimen collection semiannually to the Department. Sickle cell and MSUD treatment centers will be required to provide services to an increased number of children identified through the expanded Program. Treatment centers for galactosemia will be identified and in place prior to the commencement of statewide screening for that condition. It is anticipated that CAH will be dealt with in a similar fashion as primary congenital hypothyroidism, through referral to an endocrinologist. The laboratory with which the Department contracts will be required to perform testing for MSUD, hemoglobin disease, galactosemia and CAH, in addition to PKU and primary congenital hypothyroidism.

These regulations also generally affect all infants born in Pennsylvania, and, in particular, children born in populations at greatest risk for certain diseases (e.g. Mennonites and African-Americans).

COST AND PAPERWORK ESTIMATE

A. Cost

Statutorily mandated expansion of the Program to include testing for MSUD and sickle cell hemoglobinopathies (hemoglobin disease) will result in increased cost to the Commonwealth and, on a lesser scale, to health care providers. Annual costs of the Program are expected to increase by approximately $1,300,000 to cover testing and follow-up for MSUD, hemoglobin disease, galactosemia and CAH. This amount would be funded entirely by State funds. The total annual budget for the expanded Program includes testing of 150,000 specimens for each of the six diseases (PKU, primary congenital hypothyroidism, MSUD, hemoglobin disease, galactosemia and CAH), additional personnel, new and replacement equipment for the Bureau of Laboratories and the testing laboratory, and follow-up of children who are identified with one of the six diseases listed.

The cost to the private sector would be the cost incurred by health care providers in connection with providing the necessary follow-up to abnormal test results. The Department currently does not charge hospitals or parents for the costs of laboratory screening.

Expansion of the Program to include screening for MSUD, hemoglobin disease, galactosemia and CAH, however, will result in long-term savings as well. The total cost of screening all newborn children in Pennsylvania, including follow-up and some treatment for PKU, primary congenital hypothyroidism, MSUD and sickle cell disease is estimated at approximately $32 per child.

B. Additional Paperwork

The testing laboratory and health care providers will have additional reporting

responsibilities resulting from the addition of diseases to the list of diseases for which screening is required. The increase in paperwork requirements would be minimal, however, because the specimens necessary for screening for MSUD, hemoglobin disease, galactosemia and CAH will be collected on the same specimen collection form currently used solely for PKU and primary congenital hypothyroidism screening. Furthermore, the testing laboratory will report screening test results for the newly added diseases on the same report form currently used solely for PKU and primary congenital hypothyroidism. Under the proposed regulations, health care providers will be required to submit data regarding specimen collection to the Department semiannually. The existing regulations require only that this data be maintained. Paperwork requirements within the Department will not change significantly except to the extent that the addition of MSUD, hemoglobin disease, galactosemia and CAH will result in more instances in which follow-up of abnormal results will be required.

It should also be noted that the expanded Program for screening for sickle cell hemoglobinopathies (hemoglobin disease) and MSUD was mandated by statute in 1992, and has, in fact, been operating since that time. As has been stated, screening for sickle cell hemoglobinopathies (hemoglobin disease) began in September of 1992, and for MSUD began in March of 1993. Screening for galactosemia and CAH began on a voluntary basis in state fiscal year 2000/2001. These proposed regulations will not add to the paperwork currently being done by providers of their own volition, nor will they, for the most part, increase costs currently incurred as screening mandated by the Act is carried out.

EFFECTIVENESS/SUNSET DATES

The regulations will become effective upon final publication in the Pennsylvania Bulletin. No sunset date has been established; the Department will continually review and monitor the effectiveness of the Program.

STATUTORY AUTHORITY

The Department obtains its authority to promulgate these regulations from several sources. Generally, the Disease Prevention and Control Law of 1955 (35 P.S. §521.1 et seq.) provides the Advisory Health Board with the authority to issue rules and regulations on a variety of issues relating to communicable and non-communicable diseases, including the methods of reporting diseases, the contents of those reports and the health authorities to whom diseases are to be reported. (35 P.S. §521.16(a)). Section 16(b) of the Disease Prevention and Control Law of 1955 (35 P.S. §521.16(b)) gives the Secretary of Health (Secretary) the authority to review existing regulations and make recommendations to the Board for changes the Secretary considers to be desirable.

The Department also finds general authority for the promulgation of its regulations in the Administrative Code of 1929 (71 P.S. §51 et seq.). Section 2102(g) of the Administrative Code (71 P.S. 532(g)) gives the Department the general authority. Section 2111(b) of the Administrative Code (71 P.S. §541(b)) provides the Advisory Health Board with additional authority to promulgate regulations deemed by the Board to be necessary for the prevention of disease, and for the protection of the lives and the health of the people of the Commonwealth. That section further provides that the regulations of the Board shall become the regulations of the Department.

The Department's specific authority for promulgating the regulations relating to newborn screening and follow-up is found in the Act (35 P.S. §§621-625). Section 5 of the Act (35 P.S. §625) provides the Department, with the approval of the Board, with the authority to promulgate regulations for the implementation and administration of the Act. Section 3(b) of the Act (35 P.S. §623(b)) provides the Department, with the approval of the Board, with the authority to establish by regulation those diseases for which newborn children shall be tested and the methods for testing and disseminating test results. Section 4(b) of the Act (35 P.S. §624(b)) provides the Department with the authority to establish by regulation the methods of procurement of blood specimens of newborn children by health care providers.

REGULATORY REVIEW

Under section 5(a) of the Regulatory Review Act, the Act of June 30, 1989 (P.L. 73, No. 19), (71 P.S. §§745.1 - 745.15), the Department submitted a copy of this proposed regulation on April 18, 2001 to IRRC and to the Chairpersons of the House Health and Human Services Committee and the Senate Public Health and Welfare Committee. In addition to submitting the regulation, the Department has provided IRRC and the Committees with a copy of a detailed Regulatory Analysis Form prepared by the Department in compliance with Executive Order 1996-1, "Regulatory Review and Promulgation." A copy of this material is available to the public upon request.

If IRRC has any objection to any portion of the proposed regulation, it will notify the agency within 10 days of the close of the Committees' review period. Such notification shall specify the regulatory review criteria, which have not been met by that portion. The Act specifies detailed procedures for review, prior to final publication of the regulation by the Department, the General Assembly and the Governor, of objections raised.

CONTACT PERSON

Interested persons are invited to submit written comments, suggestions or objections regarding the proposed regulations within 30 days following publication to Jack W. Means,

Jr., Director of the Newborn Screening and Genetic Services Section of the Division of Maternal and Child Health, Bureau of Family Health, Department of Health, P.O. Box 90, Harrisburg, PA 17108, (717) 783-8143. If you are a person with a disability, comments, suggestions, or objections regarding the proposed regulations may also be submitted to Mr. Means in alternative formats, such as by audio tape, braille or using ITT (717)705-5494. If you are a person with a disability and require an alternative format of this document (e.g., large print, audio tape, braille), please contact Mr. Means so that he may make the necessary arrangements.

ANNEX A

TITLE 28. HEALTH AND SAFETY

* * *

PART III. PREVENTION OF DISEASE

* * *

CHAPTER 27. COMMUNICABLE AND NONCOMMUNICABLE DISEASES

* * *

Section 27.2. Reportable diseases.

The Board declares the following communicable diseases, unusual outbreaks of illness, noncommunicable diseases and conditions to be reportable:

* * *

Congenital adrenal hyperplasia (CAH) in children under 5 years of age.

* * *

Galactosemia in children under 5 years of age.

* * *

Maple syrup urine disease (MSUD) in children under 5 years of age.

* * *

Phenylketonuria (PKU) in children under 5 years of age.

* * *

Primary congenital hypothyroidism in children under 5 years of age.

* * *

Sickle cell disease in children under 5 years of age.

* * *

Section 27.22. Reporting laboratory results indicative of certain infections or conditions.

* * *

(b) The conditions or diseases to be reported include the following:

Congenital adrenal hyperplasia (CAH) in children under 5 years of age.

* * *

Galactosemia in children under 5 years of age.

* * *

[Hypothyroidism in infants up to 24 months old.]

* * *

Maple syrup urine disease (MSUD) in children under 5 years of age.

* * *

Phenylketonuria (PKU) in children under 5 years of age

* * *

Primary congenital hypothyroidism in children under 5 years of age.

* * *

Sickle cell disease in children under 5 years of age.

* * *

(d) The report shall be submitted by the person in charge of a laboratory as follows:

(1) *Reports except for venereal diseases, [hypothyroidism in infants up to 24 months old, phenylketonuria] CAH in children under 5 years of age, galactosemia in children under 5 years of age, MSUD in children under 5 years of age, PKU in children under 5 years of age, primary congenital hypothyroidism in children under 5 years of age, sickle cell disease in children under 5 years of age, and lead poisoning or lead toxicity.* Reports shall be made to the appropriate health authority of Philadelphia or the county department of health if the patient resides in such an area. Other reports shall be sent to the Division of Epidemiology, Department of Health, Post Office Box 90, Harrisburg, Pennsylvania 17108.

* * *

(3) [*Phenylketonuria and hypothyroidism in infants up to 24 months old*] *CAH in children under 5 years of age, galactosemia in children under 5 years of age, MSUD in children under 5 years of age, PKU in children under 5 years of age, primary congenital hypothyroidism in children under 5 years of age, and sickle cell disease in children under 5 years of age*. Reports shall be made to the Division of Maternal[/] and Child Health, Department of Health, Post Office Box 90, Harrisburg, Pennsylvania 17108.

* * *

Section 27.30. Reporting test results [of metabolic disease testing in] which identify specific diseases of the newborn [child].

In addition to the requirements that may be applicable under this chapter, testing conducted on newborn children shall be reported in accordance with Chapter 28 (relating to [metabolic] screening and followup for diseases of the newborn).

* * *

CHAPTER 28. [METABOLIC] SCREENING AND FOLLOWUP FOR DISEASES OF THE NEWBORN

GENERAL PROVISIONS

Sec.
28.1. Definitions.
28.2. [Metabolic] Newborn diseases listed.
28.3. [Tests to be performed.] Reserved.
28.4. [Standards for collecting and testing specimens.] Reserved.
28.5. Confidentiality.

[ADVICE TO PARENTS EXPLAINING] PURPOSE AND ADMINISTRATION OF TESTS

28.11. Informing the parent or guardian.
28.12. Religious objections.

[TIMING OF COLLECTION, HANDLING OF SPECIMENS AND REPORTS] SPECIMEN COLLECTION AND FOLLOWUP

28.21. Responsibility for collecting and testing initial and repeat specimens.
28.22. Timing of initial specimen collection [and handling in health care facilities] by birth centers or hospitals.
28.23. Timing of initial specimen collection [and handling for home births] by health care practitioners.
28.24. [Negative] Normal test results.
28.25. [Followup recall] Circumstances requiring repeat specimens.
28.26. Timing of [recall] repeat specimen collection[, handling and reporting].
28.27. [Followup of presumptive positive] Abnormal screening test results.
28.28. Followup of symptoms consistent with [metabolic] newborn diseases.
28.29. [Confirmatory test specimen required.] Reserved.
28.30. [Phenylketonuria] Reserved.
28.31. [Hypothyroidism] Reserved.

RECORDS

28.41. Recordkeeping requirements.

GENERAL PROVISIONS

Section 28.1. Definitions.

The following words and terms, when used in this chapter, have the following meanings, unless the context clearly indicates otherwise:

Abnormal confirmatory test result-- A test result obtained from a specimen of blood, serum, or plasma which is diagnostic of the newborn disease under investigation.

Abnormal screening test result-- A test result obtained from a specimen collected on a specimen collection form which is outside the parameters for a normal test result according to testing criteria applicable to the screening test result.

Admission-- The formal acceptance of custody or care by a [health care facility] birth center or hospital of a newborn child who is provided with bassinet or incubator, nutrition and continuous nursing service.

[*Childbearing*] *Birth center* – [A facility owned and operated by an individual, group of individuals, health agency or corporation except a hospital to provide antenatal, intrapartum and postpartum services] As defined in section 802a of Chapter 8 of the Health Care Facilities Act (35 P.S. § 448.802a).

[*Bureau*-- The Bureau of Laboratories of the Department.]

[*Confirmatory test specimen*-- A specimen of blood, serum or plasma collected from the newborn child on which a confirmatory test is performed in accordance with standards established or approved by the Bureau relating to the quantitative determination of these constituents; results of the tests may be used for diagnostic purposes.]

Days of age-- The measurement of age of the newborn child in 24-hour periods so that a newborn child is one day of age 24 hours after the hour of birth.

Department-- The Department of Health of the Commonwealth.

Discharge-- The release of the newborn child from care and custody within and by the [health care facility] birth center or hospital to the care and custody of the parent or guardian.

[*Health care facility*—A hospital or institution licensed or supervised by the Commonwealth and approved to provide impatient perinatal or pediatric services or both, and childbearing center.]

Health care practitioner-- A licensed physician or a practitioner licensed [to provide maternity care and] to deliver and care for pregnant women and newborn children.

Health care provider-- A birth center, hospital or health care practitioner.

Hemoglobin diseases-- Sickle cell (SS, SC, SV, S β Thalassemia, S O Arab) disease or trait or other clinically significant hemoglobin (CC, EE, F, H) disease or trait.

Hospital—As defined in section 802a of Chapter 8 of the Health Care Facilities Act (35 P.S. § 448.802a).

[*Hypothyroid referral clinic laboratory*-- A clinic/laboratory sponsored and supported by the Department to provide followup serum laboratory testing, consultation, diagnosis and treatment of infants with hypothyroidism.]

Inconclusive screening test result-- A test [in which the] result obtained from a specimen collected on a specimen collection form that is equivocal [by criteria established or approved by the Bureau] according to criteria applicable to the screening test result and which indicates the need for a [recall] repeat specimen and repeat testing.

[*Initial presumptive positive test*-- A test result indicating that a metabolic disease listed in section 28.2 (relating to metabolic diseases listed) may be present; the results shall be followed by confirmatory testing for diagnostic purposes.]

Initial specimen-- The first sample of blood collected [for testing purposes] from the newborn [on a special filter paper collecting device] child and submitted for testing purposes on a specimen collection form.

[*Initial test*-- The first analysis performed on an initial specimen.]

Newborn child-- An infant less than 28 days of age.

Newborn screening program--The association of the Department, the testing laboratory, and the health care provider to ensure that every newborn child born in the Commonwealth of Pennsylvania has a blood specimen collected and screened for the newborn diseases listed in §28.2 (relating to newborn diseases listed).

[*Phenylketonuria Program Clinic*-- A clinic sponsored and supported by the Department to provide expert consultation, diagnosis and treatment for children with phenylketonuria.]

Presumptive abnormal test result-- An abnormal screening test result which is sufficiently abnormal to indicate the probable presence of a newborn disease listed in §28.2.

[*Recall*] *Repeat specimen*-- A specimen collected from [the] a newborn child on a specimen collection form after the initial specimen [; such specimens are collected for the following reasons:

(i) Early discharge of newborn child from hospital.
(ii) Unacceptable specimen.
(iii) Inconclusive test result].

[*Recall*] *Repeat test*-- The laboratory [test] testing performed on a [recall] repeat specimen.

Specimen collection form-- The official newborn screening program specimen form that includes both a multipart section for providing required information about the newborn child and a filter paper tab for application of blood.

Testing laboratory—[Licensed] The licensed clinical laboratory under contract with the Department to perform [screening tests] testing for the [metabolic] newborn diseases listed in §28.2.

Transfer-- The release of the newborn child from care and custody within and by [the health care facility for] a birth center or hospital and subsequent admission to [care and custody of] another [heath care facility] hospital [or to a similar health care facility in another state].

Treatment center-- A center under contract with the Department to provide expert consultation, diagnosis, and treatment for children with a presumptive abnormal test result.

Unacceptable specimen—[Blood] A blood specimen collected from a newborn child on a [special filter paper collecting device] specimen collection form which is found to be [unacceptable] unsuitable for testing in accordance with [standards established or approved] accepted laboratory testing standards as determined by the [Bureau] Department.

Section 28.2. [Metabolic] Newborn diseases listed.

[The] A newborn child born in the Commonwealth of Pennsylvania shall be screened for the following [metabolic] diseases [of the newborn child are believed to] which may cause mental retardation [or], physical defects, or death [in the newborn child] if not detected and treated soon after birth:

(1) Congenital adrenal hyperplasia (CAH).
(2) Galactosemia.
(3) Hemoglobin diseases.
(4) Maple syrup urine disease (MSUD).
[(1)] (5) Phenylketonuria (PKU).
[(2)] (6) Primary congenital [Hypothyroidism] hypothyroidism.

Section 28.3. [Tests to be performed] (Reserved).

[(a) The following tests have been approved for the screening and detection of the these diseases:

(1) *Phenylketonuria.* The following tests are approved for detection of phenylketonuria:

(i) The Guthrie Bacterial Inhibition Assay (GBIA).

(ii) The McCaman and Robins Fluorometric method.

(iii) Other tests approved by the Bureau.

(2) *Hypothyroidism.* The following tests are approved for detection of hypothyroidism:

(i) Radioimmunoassay techniques for Thyroxine (T4) and Thyroid Stimulating Hormone (TSH) according to standards established or approved by the Bureau.

(ii) Other tests approved by the Bureau.

(b) Tests may not be administered if the parent or guardian of the newborn child objects on the grounds that the tests conflict with parent or guardian's religious beliefs or practices.]

Section 28.4. [Standards for collecting and testing specimens] (Reserved).

[A health care facility required by law or regulation to administer or cause to be administered tests for the detection of metabolic diseases in the newborn child, as specified in §28.2 (relating to metabolic diseases listed), shall collect specimens necessary to conduct the tests in accordance with standards established by the Bureau. Specimens collected shall be sent by first class mail or by other means acceptable to the Department to laboratory specified by the Department within 48 hours of collection. The Bureau will ensure the commencement of testing procedures by the testing laboratory within 48 hours of receipt of the specimen. Chapter 5 (relating to clinical laboratories), applies to the laboratory performing the tests specified in §28.3 (relating to tests to be performed).]

Section 28.5. Confidentiality.

(a) No health care provider, testing laboratory, the Department, or any other entity involved in the newborn screening program shall release any identifying information relating to any newborn child screened in the newborn screening program to anyone other than a parent or guardian of the newborn child or the health care provider for the newborn child designated by a parent or the guardian except as follows:

(1) As may be necessary to provide services to the newborn child.

(2) With the consent of the newborn child's parent or guardian.

(3) With the child's consent when the child is eighteen years of age or older, or has graduated from high school, or has married, or has been pregnant.

(b) Only the Department shall have the authority to release or authorize the release of non-identifying information concerning the newborn screening program.

[ADVICE TO PARENTS EXPLAINING] PURPOSE AND ADMINISTRATION OF TESTS

Section 28.11. Informing the parent or guardian.

[The] Prior to specimen collection, the health care [facility or practitioner responsible for care of the pregnant woman or mother] provider shall provide [her] the pregnant woman, prior to the infant's birth, or the mother or guardian, after the infant's birth, with a pamphlet supplied by the Department to explain the nature of the newborn screening [neonatal] blood tests for the [metabolic] diseases listed in §28.2 (relating to [metabolic] newborn diseases listed).

Section 28.12. Religious objections.

(a) No health care provider shall collect or cause to be collected, a specimen from a newborn child if the parent or guardian of the newborn child objects on the ground that the specimen collection conflicts with religious beliefs or practices held by the parent or guardian.

(b) If the parent or guardian of the newborn child objects to [a test] the collection of the specimen for screening on the ground that the [test] specimen collection conflicts with [his] religious beliefs or practices held by the parent or guardian, the health care [facility or practitioner responsible for the care of the newborn child shall be responsible to see] provider shall ensure that the recorded objection of the parent or guardian is entered into the medical record of the newborn child. The entry shall include a written statement of the objection signed by the parent or guardian.

[TIMING OF COLLECTION, HANDLING OF SPECIMENS, AND REPORTS] SPECIMEN COLLECTION AND FOLLOWUP

Section 28.21. Responsibility for collecting and testing initial and repeat specimens.

(a) [The health care facility or practitioner to whom care of the newborn has been entrusted or who assisted the mother at delivery shall direct blood specimens to be collected and sent for testing in accordance with §§28.4, 28.11 and 28.12 (relating to standards for collecting and testing specimens; informing the parent or

guardian; and religious objections)] A birth center or hospital shall collect or cause to be collected from each newborn child delivered in that birth center or hospital, in accordance with instructions for newborn screening specimen collection in subsection (d), the initial and any repeat specimens necessary to conduct the tests necessary for the detection of the newborn diseases specified in §28.2 (relating to newborn diseases listed).

(b) When a newborn child is delivered other than in a birth center or hospital, the health care practitioner who delivered the newborn child shall collect or cause to be collected from the newborn child, in accordance with instructions for newborn screening specimen collection in subsection (d), the initial and any repeat specimens necessary to conduct the tests necessary for the detection of the newborn diseases specified in §28.2.

(c) The health care provider shall designate a newborn screening coordinator to do the following:

(1) Ensure that a specimen collection form contains correct and complete information.

(2) Ensure that the individual who collected the specimen records that act in the newborn child's medical record.

(3) Send all specimens collected by first class mail to the testing laboratory within 24 hours of collection.

(4) Record the laboratory screening results in the newborn child's medical records.

(5) Check each newborn child's record prior to discharge or release to ensure that a specimen has been collected.

(6) Ensure, in the event of transfer of the newborn child prior to 48 hours of age, that the receiving health care provider has been notified that it has the responsibility to collect the initial specimen.

(7) Assist the Department in followup of an abnormal or presumptive abnormal test result.

(8) Followup inconclusive test results.

(9) Receive notification from the testing laboratory or from the Department of the need for a repeat specimen.

(d) The health care provider shall ensure that the individual responsible for specimen collection shall collect the specimen necessary to conduct tests in accordance with consensus standards developed by the National Committee for Clinical Laboratory Standards (NCCLS) and accepted by the Department. The Department will publish these standards, and any revisions thereto, in a notice in the *Pennsylvania Bulletin*.

Section 28.22. Timing of initial specimen collection [and handling in health care facilities] by birth centers or hospitals.

(a) [An] A birth center or hospital shall collect the initial specimen [shall be collected in health care facilities] from each newborn [infants] child, [irrespective] regardless of [age or] feeding history [,discharged on or before the fifth day of age, as close to the time of discharge from the health care facility as is practicable] or medical condition, as close to 48 hours of age as possible but not later than 72 hours of age unless the newborn child falls into one of the following categories:

(1) *Transfer.* If the newborn child is transferred to another [health care facility] hospital for continuing care [on or before the fifth day of age, the initial,] prior to 48 hours of age, the hospital to which the newborn child has been transferred shall collect a specimen [shall be collected between the fifth and sixth day of age] from the newborn child, [irrespective] regardless of feeding history [,by the health care facility to which the newborn child has been transferred] or medical condition, as close to 48 hours of age as possible but not later than 72 hours of age.

[(2) *Late discharge.* If the newborn child is discharged from the health care facility beyond the fifth day of age, the initial blood specimen shall be collected from the newborn child between the fifth and sixth day of age, irrespective of feeding history.

(3) *Instability.* If the newborn child is transferred or detained and the child's medical condition is unstable and renders the collection of the specimen undesirable at the designated time as stated in paragraph (1) or (2), whenever practicable, the initial specimen shall be collected as soon as it is deemed appropriate, but within the first 6 to 9 days of age.]

[(4)] (2) *Exchange transfusion.* [Where] If the newborn child is to undergo an exchange transfusion, the birth center or hospital shall collect the initial specimen [shall be collected] for testing immediately prior to the exchange transfusion.

[(5)] (3) *Early discharge.* [Where] If the newborn child is discharged from the [health care facility] birth center or hospital before 24 hours of age, the birth center or hospital shall collect the initial [blood] specimen [shall be collected] from the newborn child as close to the time of discharge as is practicable, [irrespective] regardless of feeding history or medical condition. [Arrangements with the parent or guardian shall be made by the health care facility or the practitioner] The birth center or hospital shall give the parent or guardian in whose care and custody the newborn child is discharged written notification of the need for a repeat specimen and shall also provide instructions to the parent or guardian for obtaining a [recall blood] repeat specimen from the newborn child as described in §28.26 (relating to timing of [recall] repeat specimen collection [, handling and reporting]).

(b) When a newborn child, who was delivered other than in a birth center or hospital, is admitted to a hospital within the first 27 days of age and the hospital has received no record of results of an approved screening test for the newborn diseases listed in §28.2 (relating to newborn diseases listed), the hospital to which the newborn child is admitted shall collect the initial specimen within 48 hours of admission to the hospital and shall send the specimen to the testing laboratory specified by the Department within 24 hours of collection.

Section 28.23. Timing of initial specimen collection [and handling for home births] by health care practitioners.

[(a) When a newborn child is born at home and is not admitted to a health care facility by the fifth day of age,] A health care practitioner who delivers a newborn child other than in a birth center or hospital shall collect or cause to be collected the initial specimen [shall be obtained] from the newborn child [and sent for testing between the second and sixth day of age by the practitioner to whom care of the newborn child has been entrusted or who assisted the mother at time of delivery or by the person who signed the newborn child's birth certificate], regardless of feeding history or medical condition, as close to 48 hours as possible but not later than 72 hours of age.

[(b) When a newborn child is admitted to a health care facility within the first 27 days of age who has not been born in nor admitted to a health care facility within the first 5 days of age and who has no record of results of an approved screening test for the metabolic diseases listed in §28.2 (relating to metabolic diseases listed), the initial specimen shall be collected within 48 hours of admission to the facility and sent for testing.]

Section 28.24. [Negative] Normal test results.

(a) No later than 7-calendar days following the day when the testing laboratory obtains the [negative] normal test results, the testing laboratory shall send those results to the health care [facility or practitioner under whose care the specimen was collected] provider that collected the specimen from the newborn child.

(b) The health care [facility or practitioner] provider to whom the [negative] normal test results are reported shall record the test results in the medical record of the [patient] newborn child.

Section 28.25. [Followup recall] Circumstances requiring repeat specimens.

(a) [If] The health care provider responsible for collecting the initial specimen shall collect or cause to be collected and submit for testing a repeat specimen if the initial specimen collected is either of the following:

(1) [unacceptable] Unacceptable for testing [if the results of testing are inconclusive, a recall specimen is required].

(2) Yields an inconclusive screening test result.

(b) If a birth center or hospital collects the initial specimen from a newborn child prior to 24 hours of age because the newborn child is discharged from the birth center or hospital prior to 24 hours of age, the birth center or hospital shall collect or cause to be collected a repeat specimen.

(c) If the initial specimen collected yields an abnormal screening test result, the Department may require the health care provider responsible for collecting the initial specimen to collect a repeat specimen.

Section 28.26. Timing of [recall] repeat specimen collection[, handling, and reporting].

(a) When the newborn child has been discharged from [the health care facility] a birth center or hospital before 24 hours of age, [a recall filter paper specimen shall be collected for testing between the sixth to ninth day whenever practicable by the health care facility or practitioner to whom care of the newborn child has been entrusted] the birth center or hospital shall collect or cause to be collected a repeat specimen from the newborn child, regardless of feeding history or medical condition, as close to 48 hours of age as possible but not later than 72 hours of age.

(b) When the initial specimen is unacceptable [for testing] or when [the results of] the initial specimen [are] yields an inconclusive screening test result, [a recall filter paper specimen shall be obtained promptly from the newborn child upon telephone notification from] the Department [to the] or testing laboratory will notify the health care [facility or practitioner who is providing ongoing care to the child according to procedures delineated in Sections 28.3(b) and 28.12 (relating to tests to be performed; and religious objections)] provider that collected the initial specimen. Within 72 hours of receipt of notice from the Department or testing laboratory, the health care provider that collected the initial specimen shall collect or cause to be collected from the newborn child a repeat specimen.

[(c) In a case where the parent or guardian has no ongoing health care provider for the newborn child, the Department will assist the parent or guardian in arranging for recall specimen collection.]

[(d)] (c) If the [appropriate] health care [facility or practitioner] provider cannot locate [the parents] a parent or guardian of the newborn child within four days of notification of need for a repeat [filter paper recall] specimen, the health care [facility or practitioner] provider shall [telephone] contact the Department [,which will assist in location of] for consultation regarding additional means for locating [the parents] a parent or guardian.

Section 28.27. [Followup of presumptive positive] Abnormal screening test results.

(a) [If the results of any filter paper test are presumptive positive, the health care facility or practitioner to whom the results were reported shall promptly notify the parents or guardian and arrange for followup and shall enter the report of the result into the patient's medical record.] When testing of the initial or repeat specimen yields an abnormal screening test result, the Department will notify the

health care provider that collected the specimen. The health care provider shall promptly notify a parent or guardian of the newborn child.

(b) If the health care [facility or practitioner] provider [to whom the presumptive positive test report was made] cannot locate the newborn child's parent or guardian within 48 hours of receiving [the report] notice from the Department, the health care [facility or practitioner] provider shall [notify] contact the Department [, which will assist in] for consultation regarding additional means for locating [the parents] a parent or guardian.

(c) The Department will assist the health care provider with and make available confirmatory testing.

(d) If the result of the confirmatory test is abnormal, the Department will assist with referral for diagnosis, treatment, and other followup services for the newborn child through designated treatment centers or clinical specialists.

Section 28.28. Followup of symptoms consistent with [metabolic] newborn diseases.

When a sick child exhibits [signs] symptoms suggestive of a [metabolic] newborn disease listed in §28.2 (relating to [metabolic]newborn diseases listed) and has not already been determined to have one of those [metabolic] newborn diseases, [a] the health care [facility or practitioner] provider to whom care of the sick child has been entrusted by the parent or guardian shall collect and submit a blood specimen for [metabolic] newborn disease testing in accordance with standard diagnostic procedures.

Section 28.29. [Confirmatory test specimen required.] (Reserved).

[If the results of any test are presumptive positive, collection of a confirmatory test specimen is required. Within 24 hours after the test results have been obtained, the Department will telephone the results to the appropriate health care facility or practitioner and follow up with a written report.]

Section 28.30. [Phenylketonuria.] (Reserved).

[(a) *Presumptive positive tests.* For presumptive positive tests the following shall apply:

(1) If the results of any test for phenylketonuria are presumptive positive, the Department will provide prompt confirmatory testing of the newborn child in accordance with standards established by the Bureau.

(2) The Department confirmatory laboratory testing of the newborn child will be completed within 24 hours of the receipt of the confirmatory test specimen or as soon as thereafter as practicable by the Department and will be reported and followed up under the same procedures set forth for presumptive positive tests in

Sections 28.27 and 28.29 (relating to followup of presumptive positive test results; and confirmatory test specimen required).

(3) The Department will telephone confirmatory test results to its designated Phenylketonuria Program Clinics.

(b) *Positive confirmatory tests.* If the results of the confirmatory tests for phenylketonuria are positive, the Department will arrange for, diagnosis, and treatment, and habititative and other followup services for the child and family in accordance with standards set or approved by the Department.]

Section 28.31. [Hypothyroidism.] (Reserved).

[(a) *Presumptive positive tests.* If the results of any hypothyroidism tests are presumptive positive, the Department will make available confirmatory laboratory testing in accordance with standards established or approved by its Bureau. Testing will be initiated within 24 hours of receipt of the specimen or as soon thereafter as is practicable by the Department's designated Hypothyroid Referral Clinics/Laboratories and will be reported and followed up under the same procedures set forth for presumptive positive tests in Sections 28.27 and 28.29 (relating to followup of presumptive test results; and confirmatory test specimen required).

(b) *Positive confirmatory tests.* If the results of any tests for neonatal hypothyroidism are positive, the Department will provide telephone or clinic consultative services through its designated Hypothyroid Referral Clinics/Laboratories in accordance with standards set or approved by the Department.]

RECORDS

Section 28.41. Recordkeeping requirements.

A health care [facility providing] provider offering maternity and newborn services shall [be required by the Department to keep data] collect and forward data semi-annually to the Department on the number of patients for whom specimens for [metabolic] newborn disease testing have been collected and the number of patients for whom such specimens have not been collected, together with the reason in each instance for the failure to collect.

* * *

PART IV. HEALTH FACILITIES

* * *

CHAPTER 501. BIRTH CENTERS

Section 501.3. Reports/contact person.

(a) The facility shall report regularly to the Department, on forms issued by the Department, statistical information that the Department may request and shall comply with the requirements for recordkeeping in § 28.41 (relating to recordkeeping requirements).

***

Section 501.49. Newborn infant care policies and procedures.

The newborn infant care policies, protocols, and procedures shall include, but not be limited to, the following:

***

(4) The birth center shall explain to the mother the purpose and nature of the screening tests for metabolic diseases, required by Chapter 28 (relating to [metabolic] screening and followup for diseases of the newborn), give her an informational pamphlet provided by the Department, inform her of her right to refuse the tests because of religious beliefs or practices, and see that the recorded written objection is entered into the medical record of the newborn child and signed by the parent or guardian, if screening is refused.

(5) The birth center shall [collect an initial filter paper blood specimen, for the detection of metabolic diseases, as close to the time of discharge from the facility as is practicable, irrespective of feeding history, unless the newborn is transferred to another health care facility for continuing care. Arrangements with the parent shall be made by the birth center, for collecting an additional blood filter paper specimen between the 2nd to 9th day of age] comply with the requirements for specimen collection, testing, and followup set forth in §§28.21--28.28.

* * *

Commonwealth of Pennsylvania

DEPARTMENT OF HEALTH

HARRISBURG

ROBERT S. ZIMMERMAN, JR., MPH
SECRETARY OF HEALTH

April 18, 2001

Mr. Robert E. Nyce
Executive Director
Independent Regulatory Review Commission
14th Floor, 333 Market Street
Harrisburg, PA 17101

Re: Department of Health Proposed Regulations No. 10-137
Newborn Disease Screening and Follow-up

Dear Mr. Nyce:

Attached are proposed regulations for review by the Commission in accordance with the Regulatory Review Act (71 P.S. §§745.1-745.15). These proposed regulations amend the Department of Health's regulations relating to newborn disease screening and follow-up (28 Pa. Code Ch. 28). These proposed regulations are being promulgated under the Newborn Child Testing Act (35 P.S. §§621-625) and the Disease Prevention and Control Law of 1955 (35 P.S. §521.1 et seq.).

Section 5(g) of the Regulatory Review Act, 71 P.S. §745.5(g), provides that the Commission shall, within 10 days after the expiration of the Standing Committee review period, notify the proposing agency of any objections to the proposed regulations. The Department expects the regulations to be published on April 28, 2001. A 30-day comment period is provided.

Section 5.1(a) of the Regulatory Review Act, 71 P.S. §745.5a(a), provides that upon completion of the agency's review of comments, the agency shall submit to the Commission a copy of the agency's response to the comments received, the names and addresses of the commentators who have requested additional information relating to the final-form regulations, and the text of the final form regulations which the agency intends to adopt.

The Department will provide the Commission within 5 days of receipt, a copy of any comment received pertaining to the proposed regulations. The Department will also provide the Commission with any assistance it requires to facilitate a thorough review of the proposed regulations. If you have any questions, please contact Deborah Griffiths, Director of the Office of Legislative Affairs at (717) 783-3985.

Sincerely,

Robert S. Zimmerman, Jr.
Secretary of Health

Enclosures

TRANSMITTAL SHEET FOR REGULATIONS SUBJECT TO THE REGULATORY REVIEW ACT

I.D. NUMBER:	10-137
SUBJECT:	Newborn Disease Screening and Follow-up
AGENCY:	Department of Health

TYPE OF REGULATION

X Proposed Regulation

Final Regulation

Final Regulation with Notice of Proposed Rulemaking Omitted

120-day Emergency Certification of the Attorney General

120-day Emergency Certification of the Governor

Delivery of Tolled Regulation
a. With Revisions b. Without Revisions

2001 APR 18 PM 3:01
REVIEW COMMISSION

FILING OF REGULATION

DATE	SIGNATURE	DESIGNATION
4/18	Lila J. Burris	HOUSE COMMITTEE ON HEALTH & HUMAN SERVICES
4/18	Mattie McKinney	
4-18	C. Magee	SENATE COMMITTEE ON PUBLIC HEALTH & WELFARE
4/18		
4/17/01	Stephen J. Hoffman	INDEPENDENT REGULATORY REVIEW COMMISSION
		ATTORNEY GENERAL
4/18/01	C. Lee Brown	LEGISLATIVE REFERENCE BUREAU

April 4, 2001