NYS Newborn Screening Panel

Effective date: 12/7/11

Pursuant to the authority vested in the Commissioner of Health by Section 2500-a of the Public Health Law, existing Section 69-1.2 of Subpart 69-1 of Title 10 (Health) of the Official Compilation of Codes, Rules and Regulations of the State of New York (NYCRR) is amended, to be effective upon publication of a Notice of Adoption in the State Register, as follows:

SUBPART 69-1
TESTING FOR PHENYLKETONURIA
AND OTHER DISEASES AND CONDITIONS
(Statutory authority: Public Health Law, sections 2500-a and 2500-f)

Section 69-1.2(b) is amended as follows:

(b) Diseases and conditions to be tested for shall include:

argininemia (ARG);

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3-hydroxy-3-methylglutaryl-CoA lyase deficiency (HMG);

[hyperammonemia/ornithinemia/citrullinemia (HHH);]

hypermethioninemia (HMET);

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propionic acidemia (PA);

severe combined immunodeficiency and other inherited T-cell deficiencies (SCID)

short-chain acyl-CoA dehydrogenase deficiency (SCADD);

tyrosinemia (TYR); and

very long-chain acyl-CoA dehydrogenase deficiency (VLCADD).

Regulatory Impact Statement Summary

Statutory Authority:

Public Health Law (PHL) Section 2500-a (a) provides statutory authority for the Commissioner of Health to designate in regulation diseases or conditions for newborn testing in accordance to the Department's mandate to prevent infant and child mortality, morbidity, and diseases and disorders of childhood.

Legislative Objectives:

In enacting PHL Section 2500-a, the Legislature intended to promote public health through mandatory screening of New York State newborns to detect those with serious but treatable neonatal conditions and to ensure their referral for medical intervention. Emerging medical treatments and the complexity of genetic testing require periodic reassessments of the benefits of newborn screening. These reassessments ensure that the New York State's Newborn Screening Program (the NYS Program) meets the legislative intent of preventing childhood diseases and disorders by early detection. This proposal, which would modify the newborn screening panel currently in regulation by adding severe combined immunodeficiency (SCID), and by deleting hyperammonemia/ornithinemia/citrullinemia (HHH), is in keeping with the legislature's public health aims of early identification and timely medical intervention for all the State's youngest citizens.

Needs and Benefits:

Severe Combined Immunodeficiency (SCID) is a primary immune deficiency, which results in the infant's failure to develop a normal immune system. The defining characteristic for SCID is a severe defect in the production and function of T-cells and/or B-cells. Affected infants are susceptible to a wide range of infections that are typically controlled by a normal immune system. If undetected and untreated, SCID typically leads to death in the first year of life. It is noteworthy that, in May of 2010, the U.S. Department of Health and Human Services (DHHS) Secretary Kathleen Sebelius added SCID to the core newborn screening panel that represents a national standard 30-test panel that states are encouraged to adopt.

The pediatric immunology community now recognizes this once-fatal disease is a disorder that can be treated and most likely cured at a reasonable cost. Early detection through screening is critical to successful treatment. Current estimates suggest that one in every 50,000 to 100,000 newborns may be affected; however, since many infants may succumb to infection before being diagnosed, the true incidence of SCID and related forms of T-cell immune deficiency may be higher. A DNA-based test for immune deficiency has been recently modified for accurate, high-throughput analyses, making possible its use for newborn screening. This test detects T-cell Receptor Gene Excision Circles or TRECs, which are produced during normal T-cell maturation but are absent or severely reduced in infants with SCID.

Immediately after confirming a SCID diagnosis, infants are started on intravenous immunoglobulins (IVIG) and antibiotics, and a donor search is initiated to perform stem cell

transplant from donor bone marrow or cord blood. SCID infants and children require IVIG for as long as they lack the ability to produce antibodies - before and often for some time after a transplant. If the transplant proves not totally corrective, IVIG may be needed for life. Alternatively, enzyme replacement therapy with bovine pegademase (PEG-ADA), an injectable medication, can be used to treat the approximately 40-percent of SCID patients with a form of the disorder characterized by a deficiency of the enzyme adenosine deaminase. This treatment is typically used only when the patient is not a candidate for the more conventional bone marrow transplant treatment.

General health care costs attributable to treatment of SCID-confirmed infants, including those related to a stem cell transplant (i.e., use of a surgical suite, stays in the neonatal intensive care unit) cannot be assessed due to large variations in charges for the professional component of specialists' and ancillary providers' services, and the scope of potentially required donor-matching services. However, overall health care costs would be reduced since early diagnosis of SCID provides the opportunity for less expensive treatments, and avoids medical complications, thereby reducing the number and average length of hospital stays, and emergency and intensive care services necessary due to recurrent infections in affected children.

If a matched, related donor cannot be found or a transplant fails, infants diagnosed with SCID typically are initially treated using IVIG as an outpatient procedure. Since IVIG only replaces the missing end product, but does not correct the deficiency in antibody production, the replacement therapy usually becomes necessary for the patient's entire lifespan. The cost of lifetime IVIG replacement therapy is estimated to be approximately

$600,000. Costs for enzyme replacement therapy for one form of SCID with PEG-ADA, which is designated as an orphan drug, are estimated at $3,800 per injection. PEG-ADA is administered by intramuscular injection twice weekly and once weekly after stabilization is reached, usually in one to three weeks. Costs for a transplant including a 1 year follow-up period are $300,000, while costs for an unscreened and undiagnosed child who does not receive early treatment can exceed $600,000.

This amendment also proposes to discontinue newborn screening for hyperammonemia/hyperornithinemia/homocitrullinemia (HHH). HHH syndrome is a rare inherited metabolic disorder that prevents the body from properly processing ammonia due to reduced enzyme activity. HHH syndrome is extremely rare; only about 50 cases are known. In 2008 and 2009, a total of 19 newborns were referred for evaluation/treatment because of elevated ornithine, a biomarker for HHH. None of these cases was confirmed as HHH. The NYS Program has seen no confirmed cases after four years of testing more than one million specimens. It is now widely recognized that levels of ornithine are not abnormal in children with the disease before five days of age, generally after a newborn screening specimen is collected.

Costs:

Costs to Private Regulated Parties:

Birthing facilities would incur no new costs related to collection and submission of blood specimens to the NYS Program, since the dried blood spot specimens now collected would also be tested for SCID. Discontinuance of screening for HHH would have no significant impact on birthing facilities or other regulated parties or stakeholders; costs

incurred from referral of SCID-positive infants would be offset by savings from no longer having to arrange for repeat specimens for or pursue referral of infants with high levels of HHH biomarker.

The NYS Program estimates that following implementation of this proposal, 125 newborns would screen positive for SCID annually statewide, with SCID being confirmed in seven of those infants.

Birthing facilities would likely incur minimal additional costs related to fulfilling their responsibilities for referral of screen-positive infants; such costs would be limited to human resources costs for less than 0.5 person-hour. Any birthing facility can calculate its specific cost impact based on its annual number of births and related expenses, and a referral rate of one infant per 2,100 births. The Department estimates that on average specialized care facilities would receive referrals of fewer than two infants per month for clinical assessment and additional testing to confirm or refute screening results.

Annual cost for arranging for SCID-related referrals for a facility at which 2,000 babies are delivered each year would range from ½ of $40 to ½ of $100, depending on whether clerical staff or nursing staff arranged for the referral, or specifically $20-50 a year. Larger birthing facilities (i.e., those with the resources to perform transplants) would not incur even these minimal costs for referral to another facility.

Cost savings from eliminating referral and follow-up to obtain repeat specimens for infants with high biomarker levels for HHH would offset approximately 10% of the costs for referral activities in response to a SCID-positive infant.

Costs for Implementation and Administration of the Rule:

Costs to State Government:

State-operated facilities providing birthing services and infant follow-up and medical care would incur costs and savings as described above for private regulated parties.

State Medicaid costs will not increase with regard to referral costs, as such costs are included in rates for delivery-related services, and are not separately reimbursed. Costs associated with treatment for SCIDS for Medicaid-eligible infants would generally be borne by the State, as most counties have already reached their cap for Medicaid liability. However, there would likely be a net savings to Medicaid since early diagnosis provides the opportunity for less expensive treatment, (on the order of $300,000) and avoids medical complications, thereby reducing the number and average length of hospital stays, and emergency and intensive care services necessary due to recurrent infections (which can exceed $600,000).

Costs to the Department:

Costs incurred by the Department's Wadsworth Center for performing SCID screening tests, providing short- and long-term follow-up, and supporting continuing research in neonatal and genetic diseases will be covered by State budget appropriations. The Program expects minimal to no additional laboratory instrumentation costs related to this proposal, since the necessary technology has already been purchased.

The Department will incur minimal administrative costs for notifying all New York State-licensed physicians, hospital chief executive officers (CEOs) and their designees, and

other affected parties, by letter informing them of a newborn screening panel expansion or, on an ongoing basis, of information regarding positive SCID screening results.

Costs to Local Government:

Local government-operated facilities providing birthing services and medical care to affected infants would incur the costs and savings described above for private regulated parties.

Local Government Mandates:

The proposed regulations impose no new mandates on any county, city, town or village government; or school, fire or other special district, unless a county, city, town or village government; or school, fire or other special district operates a facility, such as a hospital, caring for infants 28 days of age or under and, therefore, is subject to these regulations to the same extent as a private regulated party.

Paperwork:

No increase in paperwork would be attributable to activities related to specimen collection, and reporting and filing of test results. Facilities that submit newborn specimens will sustain minimal to no increases in paperwork, specifically, only that necessary to conduct and document follow-up and/or referral of infants with abnormal screening results. Educational materials for parents and health care professionals and forms will be updated to include information on SCID at minimal costs at the next printing.

Duplication:

These rules do not duplicate any other law, rule or regulation.

Alternative Approaches:

Potential delays in detection of SCID until onset of clinical symptoms would result in increased infant morbidity and mortality, and are therefore unacceptable. Given the recent recommendation by DHHS, which takes into account that treatment is available to ameliorate adverse clinical outcomes in affected infants, the Department has determined that there are no alternatives to requiring newborn screening for this condition.

Federal Standards:

The DHHS has recommended a core newborn screening panel that represents a national standard 30-test panel that states are encouraged to adopt. This core panel does not include HHH. A DHHS-commissioned Advisory Committee on Heritable Disorders of Newborns and Children recently recommended that states' newborn screening programs amend their test panels to include SCID. With the addition of SCID to its panel, the NYS Program would include all the DHHS-recommended tests.

Compliance Schedule:

The Commissioner of Health is expected to notify all New York State-licensed physicians by letter informing them of this newborn screening panel expansion. The letter

will also be distributed to hospital CEOs and their designees responsible for newborn screening, as well as to other affected parties.

The infrastructure and mechanisms for making the necessary referrals is already in place in birthing facilities. Consequently, regulated parties should be able to comply with these regulations as of their effective date.

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Regulatory Impact Statement

Statutory Authority:

Public Health Law (PHL) Section 2500-a requires institutions caring for infants 28 days of age or under to cause newborns to be tested for phenylketonuria, branched-chain ketonuria, homocystinuria, galactosemia, homozygous sickle cell disease, hypothyroidism, and other conditions to be designated by the Commissioner of Health pursuant to regulation. Specifically, PHL Section 2500-a (a) provides statutory authority for the Commissioner of Health to designate in regulation other diseases or conditions for newborn testing in accordance to the Department's mandate to prevent infant and child mortality, morbidity, and diseases and disorders of childhood. Pursuant to this authority, 44 genetic/congenital conditions and one infectious disease have been added to the newborn testing panel by regulatory amendment since initial enactment of Section 2500-a.

Legislative Objectives:

In enacting PHL Section 2500-a, the Legislature intended to promote public health through mandatory screening of New York State newborns to detect those with serious but treatable neonatal conditions and to ensure their referral for medical intervention. This proposal, which would modify the list of 44 genetic/congenital disorders and one infectious disease currently in regulation by adding severe combined immunodeficiency and other T-cell immunodeficiencies (SCID), is in keeping with the Legislature's public health aims of early identification and timely medical intervention for all the State's youngest citizens. The changing dynamics of emerging medical treatments and the complexity of genetic

testing require periodic reassessment of the state of the art in newborn screening technology and a renewed perspective on the future directions the New York State Newborn Screening Program (the NYS Program) should take to meet the legislative intent. The Department has also determined that analysis and evaluation of the primary marker (ornithine) for hyperammonemia/ hyperornithinemia/ homocitrullinemia (HHH) should be discontinued, based on recent evidence that the marker is not elevated in infants under five (5) days old, making the test not as useful in routine newborn screening as initially believed.

Needs and Benefits:

Newborn screening is a highly successful comprehensive public health program that identifies rare genetic, congenital and functional disorders; endeavors to ensure follow-up for those affected; and ensures early medical management. Data compiled from the NYS Program and other states' programs have shown that timely intervention and treatment for certain disorders can drastically improve affected infants' survival chances and quality of life, while providing the opportunity to reduce overall SCID treatment costs for a particular infant.

Severe combined immunodeficiency (SCID) is a primary immune deficiency, which results in the infant's failure to develop a normal immune system. The defining characteristic of SCID is a severe defect in the production and function of T-cells and/or B-cells. Other defects that result in immune system dysfunction, for example, DiGeorge syndrome, may be detected by the same testing method used for SCID. SCID is often

referred to as “bubble boy disease,” as it became widely known in the 1970s from media coverage of a boy with SCID who lived for 12 years in a plastic, germ-free bubble. Affected infants are susceptible to a wide range of infections that are typically controlled in those with a normal immune system. Infection sites may be the skin, the sinuses, the throat, the ears, the lungs, the brain or spinal cord, or in the urinary or intestinal tracts, and the increased vulnerability to infection may extend to repeated infections, infections that do not clear up or unusually severe infections. If undetected and untreated, SCID typically leads to death in the first year of life. It is noteworthy that in May 2010, the U.S. Department of Health and Human Services (DHHS) Secretary Kathleen Sebelius recommended adding SCID to the core newborn screening panel representing a national standard 30-test panel that states are encouraged to adopt.

The pediatric immunology medical community now recognizes that this once-fatal disease is a disorder that can be treated and most likely cured at a reasonable cost. Early detection through screening is critical to successful treatment. A survey of more than 150 patients commissioned by the Immune Deficiency Foundation found that SCID patients who were diagnosed early and treated by 3.5 months showed a 91-percent survival rate; those treated after 3.5 months had a 76-percent survival rate. Current estimates suggest that one in every 50,000 to 100,000 newborns may be affected by the disease; however, since many infants may succumb to infection before diagnosis, the true incidence of SCID and related forms of T-cell immune deficiency may be higher. Efforts to identify SCID in newborns by examining a blood smear for white blood cells have proved unreliable and expensive. A DNA-based test for immune deficiency has been recently modified to enable accurate, high-

throughput analyses, making possible its use for newborn screening. This test detects T-cell receptor gene excision circles (TREC), which are produced during normal T-cell maturation but are absent or severely reduced in infants with SCID. This new approach holds the potential to provide rapid and accurate indication of a deficiency in the immune system immediately after birth, while the infant is still protected by circulating maternal antibodies.

Immediately after confirming a SCID diagnosis, infants are placed in isolation, vaccinations are withheld and they are started on intravenous immunoglobulins (IVIG) and antibiotics, and a donor search is initiated to perform stem cell transplant from donor bone marrow or cord blood. IVIG provides antibodies that would normally be produced by healthy immune systems to help the body fight infection. IVIG is medically indicated whenever there is a known lack of antibody; therefore, IVIG could be used in all SCID-identified infants until their immune system may be reconstituted via bone marrow transplant. SCID infants and children require IVIG for as long as they lack the ability to produce antibodies - before and often for some time after a transplant. If the transplant proves not totally corrective, IVIG may be needed for life.

All forms of SCID can be treated by a bone marrow transplant. Alternatively, enzyme replacement therapy with bovine pegademase (PEG-ADA), an injectable medication, can be used to treat the approximately 40-percent of SCID patients with a form of the disorder characterized by a deficiency of the enzyme adenosine deaminase. This treatment is typically used only when the patient is not a candidate for the more conventional bone marrow transplant treatment. Lastly, there are clinical trials underway to treat the disorder through gene therapy, via insertion of a normal copy of the defective

gene into the infant's blood-forming cells. Early diagnosis provides the opportunity for less expensive treatment, (on the order of $300,000) and avoids medical complications, thereby reducing the number and average length of hospital stays, and emergency and intensive care services necessary due to recurrent infections (which can exceed $600,000).

As explained above, SCID meets established criteria applied worldwide for inclusion into newborn screening program test panels. These criteria are: the condition must be medically significant; its incidence and prevalence must represent a matter of public health concern, or it must affect a substantial number of newborns, so that the resulting cost to society for health care and lost productivity is significant; reliable assays for diagnosis of the condition must be available and suitable for large-scale population screening; and early detection of the disorder during the neonatal period must be able to allow for medical intervention effective in amelioration or prevention of medical complications, and other health consequences.

This amendment also proposes to discontinue newborn screening for hyperammonemia/ornithinemia/homocitrullinemia (HHH). HHH syndrome is a rare inherited metabolic disorder that prevents the body from properly processing ammonia due to reduced enzyme activity. Normally, the body converts ammonia into urea that can be excreted through urine. In individuals with HHH syndrome, ammonia and two amino acids (ornithine and citrulline) build up in the body. HHH syndrome is extremely rare; only about 50 cases are known to physicians, most of these cases among the French-Canadian population of Quebec Province in Canada. In fact, the NYS Program has seen no confirmed cases after four years of testing more than one million specimens. The

Massachusetts newborn screening program also reports no cases of HHH in one million specimens screened since 1999. Other entities conducting newborn testing have reported similar results. The ability of newborn screening to identify cases of HHH in newborns is questionable. It is now widely recognized that levels of the amino acid ornithine, a biomarker for HHH, are not abnormal in children with the disease before five days of age, generally after a newborn screening specimen is collected.

In 2008, abnormal ornithine values detected by the NYS Program resulted in 197 requests for repeated testing and nine referrals for evaluation/treatment because of elevated ornithine. In 2009, there were 210 requests for repeat specimens and ten referrals. None of these cases was confirmed as HHH. It is estimated that discontinuance of HHH screening would eliminate repeat testing of approximately 200 specimens each year and would reduce the number of false-positive cases referred to specialty care centers for medical evaluation. These false-positive results contribute to NYS Program expenses for repeated testing and follow-up. Elimination of screening for HHH would reduce the workload of the testing laboratory and the NYS Program follow-up unit without adverse impact to public health. Moreover, the modification would alleviate the anxiety of parents whose newborns' specimens were identified as abnormal upon screening.

Costs:

Costs to Private Regulated Parties:

Birthing facilities would incur no new costs related to collection and submission of blood specimens to the NYS Program, since the dried blood spot specimens now collected and mailed to the NYS Program for other currently available testing would also be tested

for SCID, the disorder added by this proposed amendment. Discontinuance of screening for HHH would have no adverse impact on birthing facilities or other regulated parties or stakeholders; costs incurred from referral of SCID-positive infants would be partially offset by cost savings from no longer having to arrange for repeat testing and pursue referral of infants with high levels of HHH biomarker.

The NYS Program estimates that following implementation of this proposal, 125 newborns would screen positive for SCID annually statewide. Since timing is crucial, i.e., treatment must commence early to be effective, newborns who screen positive will require immediate referral to a facility with the requisite expertise for clinical assessment and laboratory testing. The Department estimates that on average such a facility would receive referrals of fewer than two infants per month for clinical assessment and additional testing to confirm or refute screening results. Cost figures that follow are based on 125 as a high-end estimate for the maximum number of infants statewide needing immediate referral.

Birthing facilities would likely incur minimal additional costs related to fulfilling their responsibilities for referral of screen-positive infants; such costs would be limited to human resources costs for less than 0.5 person-hour per infant referred. Annual cost for arranging for SCID-related referrals for a facility at which 2,000 babies are delivered each year would range from ½ of $40 to ½ of $100, depending on whether clerical staff or nursing staff arranged for the referral, or specifically $20-50 a year. Larger birthing facilities (i.e., those with the resources to perform transplants) would not incur even these minimal costs for referral to another facility.

On average, each birthing facility can expect to refer no more than one additional

infant per year for clinical assessment and confirmatory testing as a result of this amendment's proposal to add SCID screening to the existing newborn screening panel. This increase is expected to have minimal effect on a birthing facility's workload since at present approximately 30 infants, on average, are referred by birthing facilities statewide; with the addition of SCID this number would increase by an average of one infant. Therefore, no additional staff would be required for these institutions to comply with this proposal. Any birthing facility can calculate its specific cost impact based on its annual number of births and related expenses, and a referral rate of one infant per 2,100 births.

The Department anticipates that more than 95 percent of approximately 125 referred infants will ultimately be found not to be afflicted with SCID, based on clinical assessment and laboratory tests. Cost savings from eliminating the need to arrange for repeat specimens and referral for infants with high biomarker levels for HHH would offset costs for referral activities in response to a SCID screen-positive infant.

Costs for Implementation and Administration of the Rule:

Costs to State Government:

State-operated facilities providing birthing services and infant follow-up and medical care would incur costs and savings as described above for private regulated parties.

State Medicaid costs will not increase with regard to referral costs, as such costs are included in rates for delivery-related services, and are not separately reimbursed. Costs related to treatment of Medicaid-eligible SCID-diagnosed infants would generally be borne by the State, as most counties have already reached their cap for Medicaid liability.

However, Medicaid would likely benefit from cost savings, since early diagnosis provides the opportunity for less expensive treatment (on the order of $300,000), and avoids medical complications, thereby reducing the number and average length of hospital stays, and emergency and intensive care services necessary due to recurrent infections (which can exceed $600,000).

Costs to the Department:

Costs incurred by the Department's Wadsworth Center for performing SCID screening tests, providing short- and long-term follow-up, and supporting continuing research in neonatal and genetic diseases would be covered by State budget appropriations. The Program expects minimal to no additional laboratory instrumentation costs related to this proposal, since the necessary technology – a robot to prepare DNA and set up tests, and a thermocycler to detect the TRECs – has already been purchased.

A system for follow-up and ensuring access to necessary treatment for identified infants is fully established. No additional staff would be required as a result of this proposal, provided the NYS Program's follow-up unit is at full-staffing level. The Department will incur minimal administrative costs for notifying all New York State-licensed physicians, hospital chief executive officers (CEOs) and their designees, and other affected parties, by letter informing them of a newborn screening panel expansion, or, on an ongoing basis, of information regarding positive SCID screening results.

Costs to Local Government:

Local government-operated facilities providing birthing services and medical care to affected infants would incur the costs and savings described above for private regulated parties. For those Medicaid-eligible infants, the minimal referral costs would also be absorbed by the facility as referral activities are included in the overall rate for delivery services, and are not separately reimbursed by Medicaid.

Local Government Mandates:

The proposed regulations impose no new mandates on any county, city, town or village government; or school, fire or other special district, unless a county, city, town or village government; or school, fire or other special district operates a facility, such as a hospital, caring for infants 28 days of age or under and, therefore, is subject to these regulations to the same extent as a private regulated party.

Paperwork:

No increase in paperwork would be attributable to activities related to specimen collection, and reporting and filing of test results, as the number and type of forms now used for these purposes will not change. Facilities that submit newborn specimens will sustain minimal to no increases in paperwork, specifically, only that necessary to conduct and document follow-up and/or referral of infants with abnormal screening results.

Educational materials for parents and health care professionals will be updated to include information on SCID. This revision would be accomplished with minimal costs to modify website postings and update forms and brochures at the next printing.

Duplication:

These rules do not duplicate any other law, rule or regulation.

Alternative Approaches:

Potential delays in detection of SCID until onset of clinical symptoms would result in increased infant morbidity and mortality, and are therefore unacceptable. Given the strong indication that treatment is available to ameliorate adverse clinical outcomes in affected infants, the Department has determined that there are no alternatives to requiring newborn screening for this condition.

Federal Standards:

The DHHS has recommended a core newborn screening panel that represents a national standard 30-test panel that states are encouraged to adopt. This core panel does not include HHH. A DHHS-commissioned Advisory Committee on Heritable Disorders of Newborns and Children recently recommended that states' newborn screening programs amend their test panels to include SCID. With the addition of SCID to its panel, the NYS Program would include all the DHHS-recommended tests.

Compliance Schedule:

The Commissioner of Health is expected to notify all New York State-licensed physicians by letter informing them of this newborn screening panel expansion. The letter will also be distributed to hospital chief executive officers (CEOs) and their designees responsible for newborn screening, as well as to other affected parties that are familiar with the current specimen submission and referral process, and already have the infrastructure in place to implement it. Consequently, regulated parties should be able to comply with these regulations as of their effective date.

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Regulatory Flexibility Analysis

for Small Businesses and Local Governments

Effect on Small Businesses and Local Governments:

This proposed amendment to add one new condition – an immunodeficiency disorder known as severe combined immunodeficiency (SCID) to, and to delete one condition, an inherited metabolic disorder known as hyperammonemia/ornithinemia/homocitrullinemia (HHH) from, the list of 44 genetic/congenital disorders and one infectious disease, for which every newborn in New York State must be tested, will affect hospitals, alternative birthing centers, and physician and midwifery practices operating as small businesses, or operated by local government, provided such facilities care for infants 28 days of age or under, or are required to register the birth of a child. The Department estimates that ten hospitals and one birthing center in the State meet the definition of a small business. No facility recognized as having medical expertise in clinical assessment and treatment of SCID is operated as a small business. Local governments, including the New York City Health and Hospitals Corporation, operate 21 hospitals. New York State licenses 67,790 physicians and certifies 350 licensed midwives, some of whom, specifically those in private practice, operate as small businesses. It is not possible, however, to estimate the number of these medical professionals operating an affected small business, primarily because the number of physicians involved in delivering infants cannot be ascertained.

Compliance Requirements:

The Department expects that affected facilities, and medical practices operated as small businesses or by local governments, will experience minimal additional regulatory burdens in complying with the amendment's requirements, as functions related to mandatory newborn screening are already embedded in established policies and practices of affected institutions and individuals. Activities related to collection and submission of blood specimens to the State's Newborn Screening Program will not change, since newborn dried blood spot specimens now collected and mailed to the Program for other currently performed testing would also be used for the additional test proposed by this amendment. Discontinuance of screening for HHH would have no adverse impact on birthing facilities or other regulated parties or stakeholders.

Birthing facilities and at-home birth attendants (i.e., licensed midwives) would be required to follow up infants screening positive for SCID, and assume some responsibility for referral for medical evaluation and additional testing as they do for other conditions. The anticipated increased burden is expected to have a minimal effect on the ability of small businesses or local government-operated facilities to comply, as no such facility would experience an increase of more than one to two per month in the number of infants requiring referral.

On average, each birthing facility can expect to refer no more than one additional infant per year for clinical assessment and confirmatory testing as a result of this amendment's proposal to add SCID screening to the existing newborn screening panel. This increase is expected to have minimal effect on a birthing facility's workload since at

present approximately 30 infants, on average, are referred by birthing facilities statewide; with the addition of SCID this number would increase by an average of one infant. Therefore, no additional staff would be required for these institutions to comply with this proposal.

The Department anticipates that more than 95 percent of approximately 125 referred infants will ultimately be found not to be afflicted with SCID, based on clinical assessment and laboratory tests. Cost savings from eliminating the need to arrange for repeat specimens and referral for infants with high biomarker levels for HHH would offset costs for referral activities in response to a SCID screen-positive infant.

The Department expects that regulated parties will be able to comply with these regulations as of their effective date, upon filing with the Secretary of State.

Professional Services:

No need for additional professional services is anticipated. Birthing facilities' existing professional staff are expected to be able to assume any increase in workload resulting from the Program's newborn screening for SCID and identification of screen-positive infants. Infants with positive screening tests for SCID would be referred to a facility employing a physician and other medical professionals with expertise in SCID.

Compliance Costs:

Birthing facilities operated as small businesses and by local governments, and practitioners who are small business owners (e.g., private practicing licensed midwives

who assist with at-home births) will incur no new costs related to collection and submission of blood specimens to the State Newborn Screening Program, since the dried blood spot specimens now collected and mailed to the Program for other currently available testing would also be used for the additional test proposed by this amendment. However, such facilities, and, to a lesser extent, at-home birth attendants, would likely incur minimal costs related to following up infants screening positive for SCID, primarily because the testing proposed under this regulation is expected to result in, on average, fewer than one referral per year at each of the 11 birthing facilities that are small businesses.

The NYS Program estimates that following implementation of this proposal, 125 newborns would screen positive for SCID annually statewide. Since timing is crucial, i.e., treatment must commence early to be effective, newborns who screen positive will require immediate referral to a facility with the requisite expertise for clinical assessment and laboratory testing. The Department estimates that on average such a facility would receive referrals of fewer than one infant per month for clinical assessment and additional testing to confirm or refute screening results. Cost figures that follow are based on 125 as a high-end estimate for the maximum number of infants statewide needing immediate referral.

Communicating the need for and/or arranging referral for medical evaluation of an identified infant would require less than 0.5 person-hour; no additional staff would be required. Annual cost for arranging for SCID-related referrals for a facility at which 2,000 babies are delivered each year would range from ½ of $40 to ½ of $100, depending on whether clerical staff or nursing staff arranged for the referral, or specifically $20-50 a year. Larger birthing facilities (i.e., those with the resources to perform transplants) would

not incur even these minimal costs for referral to another facility.

Economic and Technological Feasibility:

The proposed regulation would present no economic or technological difficulties to any small businesses and local governments affected by this amendment. The infrastructure for specimen collection and referrals of affected infants are already in place.

Minimizing Adverse Impact:

The Department did not consider alternate, less stringent compliance requirements, or regulatory exceptions for facilities operated as small businesses or by local government, because of the importance of the proposed testing to statewide public health. The addition of SCID to the newborn screening panel will not impose a unique burden on facilities and practitioners that are operated by a local government or as a small business. These amendments will not have an adverse impact on the ability of small businesses or local governments to comply with Department requirements for mandatory newborn screening, as full compliance would require minimal enhancements to present specimen collection, reporting, follow-up and recordkeeping practices.

Small Business and Local Government Participation:

The Program will notify all New York State-licensed physicians by letter informing them of this newborn screening panel expansion. An informational letter will also be distributed to hospital chief executive officers (CEOs) and their designees responsible for newborn screening, as well as to other affected parties. Regulated parties that are small

businesses and local governments are expected to be prepared to participate in screening and follow-up for SCID on the effective date of this amendment because the staff and infrastructure needed for specimen collection and referrals of affected infants are already in place.

Rural Area Flexibility Analysis

Types of Estimated Numbers of Rural Areas:

Rural areas are defined as counties with a population of fewer than 200,000 residents; and, for counties with a population larger than 200,000, rural areas are defined as towns with population densities of 150 or fewer persons per square mile. Forty-four counties in New York State with a population under 200,000 are classified as rural, and nine other counties include certain townships with population densities characteristic of rural areas.

This proposed amendment to eliminate one condition - hyperammonemia/ornithinemia/homocitrullinemia (HHH) - and to add one new condition – severe combined immunodeficiency (SCID) – to the list of 44 genetic/congenital disorders and one infectious disease, for which every newborn in the State must be tested, would affect hospitals, alternative birthing centers, and physician and midwifery practices located in rural areas, provided such facilities care for infants 28 days of age or under, or are required to register the birth of a child. The Department estimates that 54 hospitals and birthing centers operate in rural areas, and another 30 birthing facilities are located in counties with low-population density townships. No facility recognized as having medical expertise in clinical assessment and treatment of SCID operates in a rural area. New York State licenses 67,790 physicians and certifies 350 licensed midwives, some of whom are engaged in private practice in areas designated as rural; however, the number of

professionals practicing in rural areas cannot be estimated because licensing agencies do not maintain records of licensees' employment addresses.

Reporting, Recordkeeping and Other Compliance Requirements:

The Department expects that birthing facilities and medical practices affected by this amendment and operating in rural areas will experience minimal additional regulatory burdens in complying with the amendment's requirements, as activities related to mandatory newborn screening are already part of established policies and practices of affected institutions and individuals. Collection and submission of blood specimens to the State's Newborn Screening Program will not be altered by this amendment; the dried blood spot specimens now collected and mailed to the Program for other currently available newborn testing would also be used for the additional test proposed by this amendment. However, birthing facilities and at-home birth attendants (i.e., licensed midwives) would be required to follow up infants screening positive for SCID, and assume referral responsibility for medical evaluation and additional testing. Discontinuance of screening for HHH would have no overall impact on birthing facilities or midwives; use of human resources and costs incurred from referral of SCID-positive infants would be offset by savings from no longer having to pursue referral of infants with high levels of HHH biomarker. This requirement is expected to affect minimally the ability of rural facilities to comply, as no such facility would experience an increase of more than one to two per month in infants requiring referral. Therefore, the Department anticipates that regulated parties in rural areas will be able to comply with these regulations as of their effective date,

upon filing with the Secretary of State.

Professional Services:

No need for additional professional services is anticipated. Birthing facilities' existing professional staff are expected to be able to assume any increase in workload resulting from the Program's newborn screening for SCID and identification of screen-positive infants. Infants with a positive screening test for SCID will be referred to a facility employing a physician and other medical professionals with expertise in SCID.

Compliance Costs:

Birthing facilities operating in rural areas and practitioners in private practice in rural areas (i.e., licensed midwives who assist with at-home births) will incur no new costs related to collection and submission of blood specimens to the State's Newborn Screening Program, since the dried blood spot specimens now collected and mailed to the Program for other currently available testing would also be used for the additional test proposed by this amendment. However, such facilities and, to a lesser extent, at-home birth attendants would likely incur minimal costs related to follow-up of infants screening positive, since the proposed added testing is expected to result in no more than one additional referral per month. Communicating the need and/or arranging referral for medical evaluation of one additional identified infant would require less than 0.5 person-hour, and these tasks are expected to be able to be accomplished with existing staff. Annual cost for arranging for SCID-related referrals for a facility at which 2,000 babies are delivered each year would

range from ½ of $40 to ½ of $100, depending on whether clerical staff or nursing staff arranged for the referral, or specifically $20-50 a year. Larger birthing facilities (i.e., those with the resources to perform transplants) would not incur even these minimal costs for referral to another facility. The Department estimates that more than 95 percent of infants will be ultimately found not to be afflicted with the target condition, based on clinical assessment and additional testing.

Minimizing Adverse Impact:

The Department did not consider less stringent compliance requirements or regulatory exceptions for facilities located in rural areas because of the importance of expanded infant testing to statewide public health and welfare. The addition of SCID to the newborn screening panel will not impose a unique burden on facilities and practitioners operating in rural areas. These amendments will not have an adverse impact on the ability of regulated parties in rural areas to comply with Department requirements for mandatory newborn screening, as full compliance would entail minimal changes to present collection, reporting, follow-up and recordkeeping practices.

Rural Area Participation:

The Program will notify all New York State-licensed physicians by letter informing them of this newborn screening panel expansion. An informational letter will also be distributed to hospital chief executive officers (CEOs) and their designees responsible for newborn screening, as well as to other affected parties. Regulated parties in rural areas are

expected to be able to participate in screening and follow-up for SCID on the effective date of this amendment.

Job Impact Statement

A Job Impact Statement is not required because it is apparent, from the nature and purpose of the proposed rule, that it will not have a substantial adverse impact on jobs and employment opportunities. The amendment proposes the addition of an immune system disorder, severe combined immunodeficiency (SCID) to, and the deletion of hyperammonemia/ornithinemia/citrullinemia (HHH) from, the scope of newborn screening services provided by the Department. It is expected that no regulated parties will experience other than minimal impact on their workload, and therefore none will need to hire new personnel. Therefore, this proposed amendment carries no adverse implications for job opportunities.