Pursuant to the authority vested in the Commissioner of Health by Section 2500-a of the Public Health Law, section 69-1.2 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.

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

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

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phenylketonuria (PKU); Pompe disease;

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

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 diseases and conditions designated by the Commissioner of Health in regulation. Pursuant to PHL 2500-a, 44 genetic/congenital conditions and one infectious disease have been added to the newborn testing panel through regulation since initial enactment of this statute.

In addition, Chapter 56 of the Laws of 2013 amended PHL Section 2500-a to add a new subdivision (c) which requires the Commissioner to add, by regulation, adrenoleukodystrophy (ALD) to the list of diseases and conditions for which testing shall be performed.

Legislative Objectives:

In enacting PHL Section 2500-a, the Legislature intended to promote public health by mandating that every infant born in New York State have screening to detect serious but treatable neonatal conditions and diseases and to ensure referral for medical intervention. Chapter 56 of the Laws of 2013 amended PHL Section 2500-a to add a new subdivision (c) which requires the Commissioner to add, by regulation, ALD to the list of diseases and conditions for which testing shall be performed.

Every state in the nation provides newborn screening for their population; however, state screening panels vary. New York State maintains one of the most comprehensive screening panels in the nation. The NYS Department of Health (Department) routinely reviews its current screening panel to ensure compliance with national recommendations.

The U.S. Secretary of Health and Human Services' Discretionary Advisory Committee on Heritable Disorders in Newborns and Children (sometimes referred to herein as DACHDNC) makes recommendations on screenings to be included on states' screening panels. Based on an objective evidence report and public health impact statements, DACHDNC has recommended the inclusion of screening for Pompe disease to the uniform panel. The Department has determined that it is appropriate to also add Pompe disease to the New York State newborn screening panel.

Moreover, the proposed regulation to amend the current list of 44 genetic/congenital disorders and one infectious disease by adding ALD and Pompe disease is consistent with the Legislature's intent to improve health outcomes by supporting early identification and medical intervention for the State's youngest citizens. Innovation in medicine and scientific advances challenge our public health programs to keep step. Continual assessment and transformative perspective ensures the Department's

Newborn Screening Program maintains the legislative objectives which established the program.

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. The needs and benefits of this program are well established. Data compiled from several state programs have shown timely intervention and treatment for certain disorders drastically improves affected infants' survival chances and quality of life. Early medical intervention also provides the benefit of reducing overall lifetime treatment costs for an affected infant.

ALD, one of a group of genetic disorders called leukodystrophies, is an inherited metabolic storage disease whereby a defect in a specific enzyme results in the accumulation of very long-chain fatty acids (VLCFA) in tissues of the body, especially the brain and the adrenal glands. Ultimately the myelin sheath, an insulating membrane that surrounds nerve cells in the brain is destroyed causing neurologic problems, and the adrenal gland malfunction causes Addison's disease. ALD affects mostly males, although some women who are carriers can have milder forms of the disease.

In 2012, DACHDNC recognized ALD as a medically important disorder supported by a well-established clinical definition, screening, diagnostic and treatment protocols. Newborn screening for ALD has until recently been limited in implementation due to the lack of an accepted standard methodology. Innovative diagnostic methods combining the physical separation capabilities of liquid chromatography with the mass analysis capabilities of mass spectrometry have resulted in a powerful technique used for applications to detect conditions requiring very high sensitivity and selectivity such as ALD.

General health care costs attributable to treatment of ALD-confirmed infants, including those related to hematopoietic stem cell transplant are difficult to assess 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, it is well established that overall health care costs would be reduced as a direct result since early diagnosis of ALD provides the opportunity to avoid medical complications, reduces the number and average length of hospital stays as well as emergency and intensive care services.

Pompe disease is an inherited metabolic and potentially fatal disorder that disables the heart and skeletal muscles. The prevalence of Pompe disease was previously estimated at 1 in every 40,000 births, however sources are now estimating the prevalence may be as high as 1 in 28,000. Incidence varies among different ethnic groups. Pompe disease is caused by mutations in an enzyme-producing gene, which

is needed to break down glycogen. The severity of the disease and the age of onset are related to the degree of enzyme deficiency. With early onset of the disease, symptoms begin in the first months of life and include feeding problems, poor weight gain, enlarged heart, muscle weakness, floppiness and head lag. Respiratory difficulties are often complicated by lung infections. Most babies experience cardiac or respiratory complications before their first birthday.

Individuals with Pompe disease are best treated by a team of specialists (such as cardiologist, neurologist, and respiratory therapist) knowledgeable about the disease, who can offer care designed to manage symptoms. Scientific research on Pompe disease has led to rapid progress in understanding the biological mechanisms and properties of the enzyme. As a result, an enzyme replacement therapy (ERT) has been developed that has shown, in clinical trials with infantile-onset patients, to decrease heart size, maintain normal heart function, improve muscle function, tone, and strength, and reduce glycogen accumulation. Without ERT, the hearts of babies with infantile onset Pompe disease progressively thicken and enlarge. These babies may die before the age of one year from either cardio-respiratory failure or respiratory infection. For individuals with late onset Pompe disease, the prognosis is dependent upon the age of onset. In general, the later symptoms begin, the slower the progression of the disease.

Costs

Costs to Private Regulated Parties:

Private regulated parties affected by the proposed regulation, including; birthing facilities, hospitals, and licensed health care providers would not incur new costs or new savings related to collection and submission of blood specimens to the Department's Newborn Screening Program. The same dried blood spot specimen currently collected for the newborn screening panel would be used for ALD and Pompe disease screening.

It is anticipated that affected birth facilities, hospitals and licensed health care providers would incur minimal additional costs related to fulfilling their responsibilities to refer screen-positive infants; such costs would be limited to human resources costs for less than 0.5 hour/per week. The cost can be estimated based on annual number of births and related expenses, and a referral rate of 1 infant per 3500 births related to ALD, 1 infant per 2500 births related to Pompe disease for clinical assessment and additional testing to confirm or refute screening results.

Costs for Implementation and Administration of the Rule:

Costs to State Government:

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. Health care costs associated with medical treatment for ALD and Pompe disease for Medicaid-eligible infants would generally be borne by Medicaid, as are current

medical costs associated with care for undiagnosed infants who are Medicaid-eligible. However, there would likely be a net savings to Medicaid since early diagnosis provides the opportunity for appropriate disease-specific medical intervention. Accurate and timely diagnosis and treatment is proven to avoid medical complications for the infant, thereby eliminating unrelated testing and significantly reducing the number and average length of hospital stays, and emergency and costly intensive care services.

Costs to the New York State Department of Health:

In accordance with section 69-1.8 of Title 10 of the New York Codes, Rules and Regulations, the Department's Newborn Screening Program records diagnoses and case follow-up information submitted by health care providers and specialty care centers; maintains tracking records on identified cases; and provides educational activities and materials to parents and providers. Costs incurred by the Department's Newborn Screening Program for performing ALD and Pompe disease 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 anticipates minimal laboratory instrumentation costs as necessary technology is available in the laboratory's current equipment inventory.

A system for follow-up and ensuring access to necessary treatment for identified infants is fully established. The Department will incur minimal administrative costs for notifying all New York State-licensed physicians and midwives, hospital chief

executive officers (CEOs) and their designees, and other affected parties, by letter of the newborn screening panel expansion and providing information regarding positive ALD and Pompe disease screening results.

Costs to Local Government:

Medicaid costs incurred by counties will not increase with regard to referral costs, as such costs are included in rates for delivery-related services, and are not separately reimbursed. County Medicaid costs would not increase for Medicaid eligible infants with a Pompe disease or ALD diagnosis identified by newborn screening. There would likely be a net savings to Medicaid since early diagnosis provides the opportunity for appropriate disease-specific medical intervention. Accurate and timely diagnosis and treatment is proven to avoid medical complications for the infant, thereby eliminating unrelated testing and significantly reducing the number and average length of hospital stays, and emergency and costly intensive care services.

Local Government Mandates:

The proposed regulation imposes no new mandates on any county, city, town or village government; or school, fire or other special district.

Paperwork:

No increase in paperwork would be attributable to activities related to specimen collection. Affected parties required to 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 both ALD and Pompe disease and will be made available to the public on the Department's and Wadsworth Center's website.

Duplication:

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

Alternative Approaches:

Potential delays in detection of ALD or Pompe disease until onset of clinical symptoms could result in increased infant morbidity and mortality, and are therefore unacceptable given recent advances in laboratory diagnostic tools and medical treatment known to ameliorate adverse clinical outcomes in affected infants.

In addition, Chapter 56 of the Laws of 2013 requires the Department to add ALD to the screening panel through regulation. In light of this, the Department has determined that there are no alternatives to requiring newborn screening for ALD and Pompe disease.

Federal Standards:

DACHDNC has recommended a core newborn screening panel that represents a national standard newborn screening panel that states are encouraged to adopt.

Every state in the nation provides newborn screening for their population; however state screening panels vary. New York maintains one of the most comprehensive screening panels in the nation and works proactively to ensure valid screening tests are included in the State's screening panel in a timely manner. ALD and Pompe disease have been nominated for inclusion in the Recommended Uniform Screening Panel for state newborn screening programs.

Compliance Schedule:

The Department will notify all New York State-licensed physicians and midwives by letter 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 statewide. Consequently, regulated parties should be able to comply with the proposed regulation as of its effective date.

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Regulatory Flexibility Analysis for Small Business and Local Governments

Effect on Small Businesses and Local Governments:

The proposed regulation, which adds adrenoleukodystrophy (ALD) and Pompe disease to the list of diseases and conditions currently included on the New York State newborn screening panel, will affect hospitals, birthing centers, and physician and midwifery practices operating as small businesses, provided such facilities care for infants 28 days of age or under, or are required to register the birth of a child. However, the impact is minimal as the same dried blood spot specimen currently collected for the newborn screening panel would be used for ALD and Pompe disease screening. It is anticipated that regulated parties would incur minimal additional costs related to fulfilling their responsibilities to refer screen-positive infants, and for clinical assessment and additional testing to confirm or refute screening results.

New York State licenses approximately 82,000 physicians and 1,000 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. The Department estimates that three hospitals and one birthing center in the State meet the definition of a small business. The addition of ALD and Pompe disease to the Newborn Screening Panel will not place economic or regulatory burden on affected small parties.

Effects on local governments concern costs related to referral and treatment for affected infants. Medicaid costs incurred by counties will not increase with regard to referral costs, as such costs are included in rates for delivery-related services, and are not separately reimbursed. County Medicaid costs would not increase for Medicaid eligible infants with a Pompe disease or ALD diagnosis identified by newborn screening. There would likely be a net savings to Medicaid since early diagnosis provides the opportunity for appropriate disease-specific medical intervention. Accurate and timely diagnosis and treatment is proven to avoid medical complications for the infant, thereby eliminating unrelated testing and significantly reducing the number and average length of hospital stays, and emergency and costly intensive care services.

Compliance Requirements:

The Department expects that hospitals, birthing centers, and physician and midwifery practices operating as small businesses will not experience 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 Department'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 be used for ALD and Pompe disease screening.

Affected facilities and licensed health professionals would be required to refer infants screening positive for ALD or Pompe disease for medical evaluation, and additional testing and provide follow-up as they currently do for other conditions.

The anticipated increased burden will have a minimal effect on the ability of small businesses to comply. On average, hospitals, birthing centers, and physician and midwifery practices operating as small businesses, can expect to refer one to two additional infants per month for clinical assessment and confirmatory testing as a result of the proposed regulation. This increase is expected to have minimal effect on affected facilities and licensed health care providers' workload. It is anticipated that additional staffing resources are not required to comply with the proposed regulation.

The Department expects that regulated parties will be able to comply with the proposed regulation as of its effective date.

Professional Services:

No need for additional professional services is anticipated. Professional staff of regulated parties affected by the proposed regulation will assume any increase in workload resulting from the screening for ALD and Pompe disease and identification of screen-positive infants. Infants with positive screening tests would be referred to a facility employing a physician and other medical professionals with expertise in the respective disorder.

Compliance Costs:

Hospitals, birthing centers, and physician and midwifery practices operating as small businesses will not incur additional costs related to collection and submission of blood specimens to the Department's Newborn Screening Program, since the dried blood spot specimens now collected and mailed to the Program for the screening program currently operating would also be used to screen for the additional disorders 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, primarily because the testing proposed under this regulation is expected to result in, on average, 1 to 2 referrals per month.

Economic and Technological Feasibility:

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

Minimizing Adverse Impact:

The addition of 2 disorders to the newborn screening panel will not impose a unique burden on hospitals, birthing centers, and physician and midwifery practices operating as small businesses. The proposed regulation will not have an adverse impact on the ability of small businesses to comply with statutory 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 Department will notify all New York State-licensed physicians and midwives 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 are expected to comply with screening and follow-up for ALD and Pompe disease on the effective date of the proposed regulation because the staff and infrastructure needed for specimen collection and referrals of affected infants are already in place. Small businesses and local governments will have the opportunity to participate in the rulemaking process by submitting comments during the public comment period following the publication of the Notice of Proposed Rulemaking.

Rural Area Flexibility Analysis

Effect on Rural Areas:

Rural areas are defined as counties with a population less than 200,000 and, for counties with a population greater than 200,000, include towns with population densities of 150 persons or less per square mile. Forty three counties in New York State have a population less than 200,000, and nine other counties have certain townships with population densities of 150 persons or less per square mile.

The proposed regulation adds two disorders, adrenoleukodystrophy (ALD) and Pompe disease, to the list of genetic/congenital disorders for which every newborn in New York State must be tested. It will affect hospitals, alternative birthing centers and physician and midwifery practices located in rural areas to the extent that such facilities care for infants 28 days of age or under, or are required to register the birth of a child. Such facilities will not incur new costs related to collection and submission of blood specimens, as the same blood spot specimen that is currently collected would be used for ALD and Pompe disease screening. Facilities and practices may incur minimal additional costs related to fulfilling their responsibility to refer screen-positive infants.

Compliance Requirements:

The infrastructure to support the Newborn Screening Program is currently operational. The proposed regulation adds 2 genetic disorders to the current panel of

disorders currently screened. Minimal reporting, record keeping, or other compliance requirements are being imposed as a result of the proposed regulation.

Professional Services:

No new additional professional services are required in order for providers in rural areas to comply with the proposed regulation.

Compliance Costs:

No initial capital costs will be imposed as a result of the proposed regulation, nor is there an annual cost of compliance.

Minimizing Adverse Impact:

New York State's Newborn Screening program was first implemented in 1965 and screened for a single metabolic disorder. With almost 50 years of experience, the program now conducts more than 12 million screens to identify congenital disorders and exposure to HIV on 250,000 babies annually. The proposed regulation expands diagnostic opportunity and improves health outcomes without increased burden to providers located in rural areas or otherwise. The goal is to ensure this highly successful, comprehensive public health program is made available to all newborns across New York.

The proposed regulation will not have an adverse impact on the ability of hospitals, alternative birthing centers and physician and midwifery practices located in rural

areas to comply with statutory requirements for mandatory newborn screening, as full compliance would require no change to the collection of blood spot specimens as the same dried blood spot specimen currently collected for the newborn screening panel will be used for ALD and Pompe disease screening. The proposed regulation will require minimal enhancements to reporting, follow-up and recordkeeping practices. The minimal increase in workload attributed to referring affected infants is expected to be assumed by current personnel.

Opportunity for Rural Area Participation:

The proposed regulation adds two additional disorders to the list of disorders currently screened. It will not have an adverse impact on hospitals, alternative birthing centers and physician and midwifery practices located in rural areas, as implementation of an expanded disease screening panel will result in minimal new reporting, record keeping, or other compliance requirements. Public and private interests in rural areas will have the opportunity to participate in the rulemaking process by submitting comments during the public comment period following the publication of the Notice of Proposed Rulemaking.

Job Impact Statement

A Job Impact Statement for these amendments is not being submitted because it is apparent from the nature and purposes of the amendments that they will not have a substantial adverse impact on jobs and/or employment opportunities.