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Proposed Regulation
Agency Background Document

Agency name	Virginia Department of Health
Virginia Administrative Code (VAC) Chapter citation(s)	12 VAC5-71
VAC Chapter title(s)	Regulations Governing Virginia Newborn Screening Services
Action title	Amend regulations to add SMA and X-ALD to the Virginia Newborn Screening System core panel of heritable disorders and genetic diseases.
Date this document prepared	May 12, 2020

This information is required for executive branch review and the Virginia Registrar of Regulations, pursuant to the Virginia Administrative Process Act (APA), Executive Order 14 (as amended, July 16, 2018), the Regulations for Filing and Publishing Agency Regulations (1VAC7-10), and the *Form and Style Requirements for the Virginia Register of Regulations and Virginia Administrative Code*.

Brief Summary

Provide a brief summary (preferably no more than 2 or 3 paragraphs) of this regulatory change (i.e., new regulation, amendments to an existing regulation, or repeal of an existing regulation). Alert the reader to all substantive matters. If applicable, generally describe the existing regulation.

The proposed regulatory action would amend the existing newborn screening regulation to add spinal muscular atrophy (SMA) and X-linked adrenoleukodystrophy (X-ALD) to the newborn screening panel. Blood spot newborn screening services are provided by the Department of General Services' Division of Consolidated Laboratory Services (DCLS) in partnership with the Virginia Department of Health (VDH). SMA is a genetic disorder that is estimated to occur in approximately 9.1 out of every 100,000 live births. X-ALD is a genetic disorder that is estimated to occur in approximately 6 out of every 100,000 live births. Treatment for both X-ALD and SMA is available if detected early. Screening is necessary, as these disorders cannot be detected at birth through physical examinations. The additions of SMA and X-ALD to the newborn screening panel have been recommended by the Virginia Genetics Advisory Committee. On the national level, these disorders have been added to the core panel of 35 genetic disorders included in the Recommended Uniform Screening Panel (RUSP) of the U.S. Secretary of Health and Human Services' (HHS) Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC).

Acronyms and Definitions

Define all acronyms used in this form, and any technical terms that are not also defined in the "Definitions" section of the regulation.

ACHDNC – Advisory Committee on Heritable Disorders in Newborns and Children

DCLS – Division of Consolidated Laboratory Services

HHS – Health and Human Services

RUSP – Recommended Uniform Screening Panel

SMA – spinal muscular atrophy

VDH – Virginia Department of Health

VNSP – Virginia Newborn Screening Program

X-ALD – X-linked adrenoleukodystrophy

Mandate and Impetus

Identify the mandate for this regulatory change and any other impetus that specifically prompted its initiation (e.g., new or modified mandate, petition for rulemaking, periodic review, or board decision). For purposes of executive branch review, "mandate" has the same meaning as defined in Executive Order 14 (as amended, July 16, 2018), "a directive from the General Assembly, the federal government, or a court that requires that a regulation be promulgated, amended, or repealed in whole or part."

The State Board of Health is initiating this regulatory action in response to a recommendation received from the Virginia Genetics Advisory Committee. On the national level, these disorders have been added to the core panel of 35 genetic disorders included in the RUSP of the U.S. Secretary of HHS ACHDNC.

Legal Basis

Identify (1) the promulgating agency, and (2) the state and/or federal legal authority for the regulatory change, including the most relevant citations to the Code of Virginia and Acts of Assembly chapter number(s), if applicable. Your citation must include a specific provision, if any, authorizing the promulgating agency to regulate this specific subject or program, as well as a reference to the agency's overall regulatory authority.

The State Board of Health is authorized to make, adopt, promulgate and enforce regulations by Section 32.1-12 of the Code of Virginia.

Section 32.1-65 of the Code of Virginia requires newborn screening to be conducted on every infant born in the Commonwealth of Virginia.

Section 32.1-67 of the Code of Virginia requires the Board of Health to promulgate regulations as necessary to implement Newborn Screening Services. The regulations are required to include a list of newborn screening tests pursuant to Section 32.1-65.

Purpose

Explain the need for the regulatory change, including a description of: (1) the rationale or justification, (2) the specific reasons the regulatory change is essential to protect the health, safety or welfare of citizens, and (3) the goals of the regulatory change and the problems it's intended to solve.

Spinal muscular atrophy is a genetic disorder characterized by weakness and wasting (atrophy) in muscles used for movement (skeletal muscles). SMA is caused by a loss of specialized nerve cells, called motor neurons, which control muscle movement. SMA affects 9.1 out of every 100,000 births and there are five classification types. Type 0 often leads to fetal loss or newborns with significant involvement and death in early infancy; this is the rarest and most severe form of the condition. Type I, the most common form, leads to progressive weakness in the first six months of life and, without targeted intervention, death prior to two years of age. Type II is associated with progressive weakness by 15 months of life and, without targeted intervention, respiratory failure and death after the third decade of life. Types III and IV are associated with progressive weakness that develops after one year of life or in adulthood, and most individuals have a normal lifespan. Treatment for SMA generally includes a disease-modifying therapy that uses FDA-approved Spinraza, as well as clinical care support therapies such as nutritional support, respiratory support, pulmonary care, orthopedic and rehabilitation care, and palliative care.

X-linked adrenoleukodystrophy is a genetic disorder that occurs primarily in males, mainly affecting the nervous system and the adrenal glands. In the United States, X-ALD affects 6 out of every 100,000 births, regardless of sex. There are three distinct types of X-ALD: a childhood cerebral form, an adrenomyeloneuropathy type, and a form called Addison disease only. Childhood cerebral X-ALD is the most serious form of X-ALD and it usually presents between 2.5 and 10 years of age. It is associated with rapid neurologic decline and death or disability an average three years after onset. Signs and symptoms of the adrenomyeloneuropathy type appear between early adulthood and middle age. People with X-ALD whose only symptom is adrenocortical insufficiency are said to have the Addison disease only form, which is the mildest form of the three types. In these individuals, adrenocortical insufficiency can begin anytime between childhood and adulthood. Treatment for X-ALD is difficult to predict since symptom onset varies and, in many cases, might not occur until after infancy. Treatment options include hormone therapy and hematopoietic stem cell transplantation (HSCT), depending on the severity of the disorder.

All newborns in Virginia would be screened for SMA and X-ALD as a result of this proposed regulatory action. Screening for SMA and X-ALD can provide affected infants the benefit of early diagnosis and treatment. Screening is an effective diagnostic tool since these disorders cannot be detected at birth through a physical examination. Laboratory screening is available at a cost.

The addition of SMA and X-ALD to the core panel will result in an increase to the newborn screening fee. The VDH Office of Family Health Services has a longstanding partnership with DCLS to provide blood spot newborn screening services. The Virginia Newborn Screening Program is solely funded through Enterprise Funding, which is generated from the collection of fees from dried blood spot specimen kits sold to submitting birthing facilities and health care providers statewide. As of October 1, 2019, the newborn screening fee is $138 per card. To implement these two screenings statewide, DCLS will require infrastructure investment that includes additional laboratory equipment; programmatic staff; application development to incorporate screening results; incorporation of new education modules; identification of specialized medical support systems for infants and their families; and family support and case management services for infants diagnosed with SMA or X-ALD. Adding SMA to the newborn screening panel results in an increase of $2.16, and adding X-ALD to the newborn screening results in an increase of $10.84 per sample, for a total of $13 for both of these disorders.

Substance

Briefly identify and explain the new substantive provisions, the substantive changes to existing sections, or both. A more detailed discussion is provided in the "Detail of Changes" section below.

The proposed changes to 12 VAC 5-71 will revise Section 30, which lists the specific disorders and genetic diseases that must be screened in Virginia, by adding SMA and X-ALD to the state's core panel. Currently, DCLS analyzes biological markers that may be indicative of 31 certain disorders that constitute the core panel. Section 32.1-67 of the Code of Virginia requires that this list of screened disorders be in the regulation. Section 32.1-65 of the Code requires that Virginia's screening tests are consistent with the panel recommended by the U.S. Secretary of HHS ACHDNC.

Issues

Identify the issues associated with the regulatory change, including: 1) the primary advantages and disadvantages to the public, such as individual private citizens or businesses, of implementing the new or amended provisions; 2) the primary advantages and disadvantages to the agency or the Commonwealth; and 3) other pertinent matters of interest to the regulated community, government officials, and the public. If there are no disadvantages to the public or the Commonwealth, include a specific statement to that effect.

The primary advantage of the proposed regulatory action to the public is that screening for SMA and X-ALD can provide affected infants the benefit of early diagnosis and treatment. Screening is an effective diagnostic tool since these disorders cannot be detected at birth through a physical examination. The primary disadvantage to the public is that adding these two screenings to the panel results in a cost increase.

A primary advantage of the proposed regulatory action to the agency is that the action aligns with the recommendation from the Virginia Genetics Advisory Committee to add SMA and X-ALD to the state's core panel. This also aligns with the panel recommended by the U.S. Secretary of HHS ACHDNC.

A disadvantage to the regulated community, government officials and the public is the projected increase in the cost of the two screenings. Newborn screening is a fee-for-service program, and the fee is paid by hospitals and other screeners who must purchase the filter paper kits used for blood spot collection. Most screening is performed in hospitals, with about 10-15% of screening performed by private physicians and military facilities. Hospitals do not generally pass on these costs to patients because third party payers usually pay a negotiated bundled amount per delivery, and Medicaid reimbursed delivery payment is set by the state. Self-pay patients may be responsible to pay the screening fee themselves if they have the resources to do so.

Requirements More Restrictive than Federal

Identify and describe any requirement of the regulatory change which is more restrictive than applicable federal requirements. Include a specific citation for each applicable federal requirement, and a rationale for the need for the more restrictive requirements. If there are no applicable federal requirements, or no requirements that exceed applicable federal requirements, include a specific statement to that effect.

There are no requirements of this proposal that are more restrictive than federal requirements.

Agencies, Localities, and Other Entities Particularly Affected

Identify any other state agencies, localities, or other entities particularly affected by the regulatory change. "Particularly affected" are those that are likely to bear any identified disproportionate material impact which would not be experienced by other agencies, localities, or entities. "Locality" can refer to either local governments or the locations in the Commonwealth where the activities relevant to the regulation or regulatory change are most likely to occur. If no agency, locality, or entity is particularly affected, include a specific statement to that effect.

Other State Agencies Particularly Affected

The Department of General Services' DCLS is particularly affected by this regulatory change. The Department of Medical Assistance Services may also be affected since they may have to negotiate new reimbursement rates for the increased fee.

Localities Particularly Affected

No locality will be particularly affected by the proposed amendment.

Other Entities Particularly Affected

Hospitals, birthing centers and regional genetic centers within the Commonwealth will be affected by the proposed amendment.

Economic Impact

Pursuant to § 2.2-4007.04 of the Code of Virginia, identify all specific economic impacts (costs and/or benefits), anticipated to result from the regulatory change. When describing a particular economic impact, specify which new requirement or change in requirement creates the anticipated economic impact. Keep in mind that this is change versus the status quo.

Impact on State Agencies

For your agency: projected costs, savings, fees or revenues resulting from the regulatory change, including: a) fund source / fund detail; b) delineation of one-time versus on-going expenditures; and c) whether any costs or revenue loss can be absorbed within existing resources	VDH costs are included in the newborn screening fee, which include one full-time employee for follow-up activities and education and outreach costs.
For other state agencies: projected costs, savings, fees or revenues resulting from the regulatory change, including a delineation of one-time versus on-going expenditures.	Projected costs to add SMA and X-ALD to the newborn screening panel will be incurred by DCLS. Costs related to capital equipment, staff, application development and education modules to conduct SMA screenings are estimated at $389,631 start-up costs and $192,262 annually. Costs related to capital equipment, staff, application development and education modules to conduct X-ALD screenings are estimated at $1,101,568 start-up costs and $1,073,422 annually. The projected costs will be funded through the fee increase for the blood spot screening panel resulting from the addition of SMA and X-ALD to the core panel.

For all agencies: Benefits the regulatory change is designed to produce.	SMA and X-ALD are genetic disorders affecting newborns that can result in death if not treated early. These amendments will assure that all newborns born in Virginia hospitals and birthing centers will be screened for SMA and X-ALD prior to their discharge. Better health outcomes and higher infant survival rates are the intended impacts.

Impact on Localities

Projected costs, savings, fees or revenues resulting from the regulatory change.	There is no projected fiscal impact on localities.
Benefits the regulatory change is designed to produce.	These amendments will assure that all newborns born in Virginia hospitals and birth centers will be screened for SMA and X-ALD prior to their discharge. Better health outcomes and higher infant survival rates are the intended impacts.

Impact on Other Entities

Description of the individuals, businesses, or other entities likely to be affected by the regulatory change. If no other entities will be affected, include a specific statement to that effect.	Hospitals, birthing centers, midwives and infants born in Virginia hospitals and birth centers will likely be affected.
Agency's best estimate of the number of such entities that will be affected. Include an estimate of the number of small businesses affected. Small business means a business entity, including its affiliates, that: a) is independently owned and operated and; b) employs fewer than 500 full-time employees or has gross annual sales of less than $6 million.	Hospitals: 58 Birth centers: Approximately 10-15 Midwives: Unknown Infants born in these facilities annually: 99,000
All projected costs for affected individuals, businesses, or other entities resulting from the regulatory change. Be specific and include all costs including, but not limited to: a) projected reporting, recordkeeping, and other administrative costs required for compliance by small businesses; b) specify any costs related to the development of real estate for commercial or residential purposes that are a consequence of the regulatory change; c) fees; d) purchases of equipment or services; and e) time required to comply with the requirements.	The current cost of the newborn screening panel is $138. Adding SMA to the newborn screening panel results in an increase of $2.16, and adding X-ALD to the newborn screening panel results in an increase of $10.84 per sample. a) $0 b) $0 c) $13.00 increase per sample d) Start-up equipment cost is $389,631 for SMA and $1,101,568 for X-ALD. e) The fee increase needs to go into effect 12 months prior to implementation to accrue start-up costs.
Benefits the regulatory change is designed to produce.	SMA and X-ALD are genetic disorders affecting newborns that can result in death if not treated early. These amendments will assure that all newborns born in Virginia hospitals and birth centers will be screened for SMA and X-ALD prior to their discharge. Better health outcomes and higher infant survival rates are the intended impacts.

Alternatives to Regulation

Describe any viable alternatives to the regulatory change that were considered, and the rationale used by the agency to select the least burdensome or intrusive alternative that meets the essential purpose of the regulatory change. Also, include discussion of less intrusive or less costly alternatives for small businesses, as defined in § 2.2-4007.1 of the Code of Virginia, of achieving the purpose of the regulatory change.

The alternative to this proposed regulatory action is to not add SMA and X-ALD to the core panel of disorders for which newborns are screened. However, this option would be in direct conflict with both the national RUSP and the recommendation of the Virginia Genetics Advisory Committee.

Regulatory Flexibility Analysis

Pursuant to § 2.2-4007.1B of the Code of Virginia, describe the agency's analysis of alternative regulatory methods, consistent with health, safety, environmental, and economic welfare, that will accomplish the objectives of applicable law while minimizing the adverse impact on small business. Alternative regulatory methods include, at a minimum: 1) establishing less stringent compliance or reporting requirements; 2) establishing less stringent schedules or deadlines for compliance or reporting requirements; 3) consolidation or simplification of compliance or reporting requirements; 4) establishing performance standards for small businesses to replace design or operational standards required in the proposed regulation; and 5) the exemption of small businesses from all or any part of the requirements contained in the regulatory change.

VDH staff convened SMA and X-ALD workgroups comprised of internal and external stakeholders including medical experts in the field of pediatric SMA and X-ALD diagnosis and treatment, professionals from major medical and higher education institutions within the Commonwealth, parent advocates and staff from DCLS to evaluate and consider this regulatory change and its cost effectiveness. The alternative regulatory methods are not applicable. There are no other applicable regulations to consolidate which impact newborn screening. Small businesses may not be exempted as a category because screening for all infants must be managed equitably by their providers, regardless of business size, to assure optimal outcomes. There are no viable alternatives to the proposed regulatory action to achieve the necessary regulatory changes determined to be appropriate.

Periodic Review and Small Business Impact Review Report of Findings

If you are using this form to report the result of a periodic review/small business impact review that is being conducted as part of this regulatory action, and was announced during the NOIRA stage, indicate whether the regulatory change meets the criteria set out in Executive Order 14 (as amended, July 16, 2018), e.g., is necessary for the protection of public health, safety, and welfare; minimizes the economic impact on small businesses consistent with the stated objectives of applicable law; and is clearly written and easily understandable.

In addition, as required by § 2.2-4007.1 E and F of the Code of Virginia, discuss the agency's consideration of: (1) the continued need for the regulation; (2) the nature of complaints or comments received concerning the regulation; (3) the complexity of the regulation; (4) the extent to the which the

regulation overlaps, duplicates, or conflicts with federal or state law or regulation; and (5) the length of time since the regulation has been evaluated or the degree to which technology, economic conditions, or other factors have changed in the area affected by the regulation. Also, discuss why the agency's decision, consistent with applicable law, will minimize the economic impact of regulations on small businesses.

The regulation meets the criteria set out in Executive Order 14 and is necessary for the protection of public health, safety and welfare.

There is a continued need for the regulations, as the provision of newborn screening services to babies born in the Commonwealth of Virginia are required by legislation. The amendment to add SMA and X-ALD to the Virginia Newborn Screening System's core panel of heritable disorders and genetic diseases is consistent with the RUSP. SMA and X-ALD were recommended to be added to the RUSP in July 2018 and February 2016, respectively.

Two public comments were received in June 2019 in support of adding SMA to the newborn screening panel. The comments were received during the public comment period.

The regulations are clearly written and easily understandable. The regulations do not overlap, duplicate or conflict with any known federal or state law or regulation. Regulations are evaluated on an ongoing basis and these regulations were last amended in January 2019.

Public Comment

Summarize all comments received during the public comment period following the publication of the previous stage, and provide the agency response. Include all comments submitted: including those received on Town Hall, in a public hearing, or submitted directly to the agency. If no comment was received, enter a specific statement to that effect.

Commenter	Comment	Agency response
Debra Schaefer, Virginia Chapter Cure SMA	I can't express strongly enough the importance and urgency of adding Spinal Muscular Atrophy (SMA) to Virginia's Newborn Screening Panel as expeditiously as possible. My family has experienced the heartbreaking loss of my first granddaughter 7 years ago to SMA, at the tender age of 7 months. At that time, there was no treatment and no cure. We are now experiencing the joy of seeing my second granddaughter live and thrive with SMA since her participation from the time she was 3 months of age in the clinical trial that resulted in the first FDA approved treatment for SMA. She had already lost most of her ability to move. Although still medically fragile, she is 5 1/2 years old and able to do things previously unheard of for a Type 1 baby, including holding up her head and propelling her own manual wheelchair. We have seen firsthand the profoundly improved outcomes for the Type 1 babies who are diagnosed and treated PRIOR to exhibiting symptoms. PLEASE expedite implementation of Newborn Screening for SMA in Virginia so that approximately 9 families per year will never know the struggles we will face every day.	VDH notes the support of the proposed amendments. No response is required.

Jaimie Vickery, Cure SMA	On behalf of the largest nonprofit organization dedicated to finding a cure for spinal muscular atrophy (SMA), we ask that Virginia adopt newborn screening for SMA as soon as possible. SMA is the most common genetic cause of death in infants in the United States, affecting approximately 1 in 11,000 newborns. The condition is caused by a mutation in the survival motor neuron gene 1 (*SMN1*) that causes nerve cells to malfunction, leading to debilitating and often fatal muscle weakness. In Virginia, 9 babies are born with SMA every year, and roughly 155,000 individuals are genetic carriers of the condition. Fortunately, there are two FDA-approved treatments for the disease, but they cannot repair motor neuron damage that has already happened, only slow down or prevent further damage. Because of this, treatment must happen as soon as possible for it to be most effective. In some cases, this may be before a child shows any symptoms of the disease. It is critical, therefore, that newborns with SMA be identified and receive treatment as soon as possible. Given the importance of newborn screening in effectively treating SMA, Health and Human Services Secretary Alex Azar added SMA to the Recommended Uniform Screening Panel in July of 2018, and Virginia's Advisory Council voted to add it in November 2018. More than twenty other states have approved adding SMA to their newborn testing program, and seven states have already begun testing. Already, several infants have been identified and are receiving life-saving treatment. Therefore, we ask that Virginia adopt this screening as soon as possible	VDH notes the support of the proposed amendments. No response is required.

Public Participation

Indicate how the public should contact the agency to submit comments on this regulation, and whether a public hearing will be held, by completing the text below.

The Virginia Department of Health is providing an opportunity for comments on this regulatory proposal, including but not limited to (i) the costs and benefits of the regulatory proposal, (ii) any alternative approaches, (iii) the potential impacts of the regulation, and (iv) the agency's regulatory flexibility analysis stated in that section of this background document.

Anyone wishing to submit written comments for the public comment file may do so through the Public Comment Forums feature of the Virginia Regulatory Town Hall web site at: https://townhall.virginia.gov. Comments may also be submitted by mail, email or fax to Robin

Buskey, Virginia Department of Health, 109 Governor Street, Richmond, Virginia 23219, robin.buskey@vdh.virginia.gov, (804) 864-7652. In order to be considered, comments must be received by 11:59 pm on the last day of the public comment period.

A public hearing will not be held following the publication of this stage of this regulatory action.

Detail of Changes

List all regulatory changes and the consequences of the changes. Explain the new requirements and what they mean rather than merely quoting the text of the regulation. For example, describe the intent of the language and the expected impact. Describe the difference between existing requirement(s) and/or agency practice(s) and what is being proposed in this regulatory change. Use all tables that apply, but delete inapplicable tables.

Changes to Existing VAC Chapter(s)

Current section number	New section number, if applicable	Current requirement	Change, intent, rationale, and likely impact of new requirements
12VAC5-71-30		The Virginia Newborn Screening System's core panel of heritable disorders and genetic diseases.	This section lists the conditions of the core panel of heritable disorders and genetic diseases for which the newborn dried blood spot testing is conducted. The proposed change would add SMA and X-ALD to the core panel. Intent: Align Virginia Newborn screening panel with the recommendations of the Virginia Genetics Advisory Committee and the U.S. Secretary of HHS ACHDNC. Rationale: Screening for these two additional disorders can provide affected infants the benefit of early diagnosis and treatment. Likely Impact: Better health outcomes and higher infant survival rates.