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Final Regulation
Agency Background Document

Agency name	Virginia Department of Health
Virginia Administrative Code (VAC) Chapter citation(s)	12VAC5-71
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	7/12/2021

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

Statement of Final Agency Action

Provide a statement of the final action taken by the agency including: 1) the date the action was taken; 2) the name of the agency taking the action; and 3) the title of the regulation.

The State Board of Health approved these final amendments to the Regulations Governing Virginia Newborn Screening Services on December 10, 2021.

Mandate and Impetus

List all changes to the information reported on the Agency Background Document submitted for the previous stage regarding the mandate for this regulatory change, and any other impetus that specifically prompted its initiation. If there are no changes to previously reported information, include a specific statement to that effect.

There are no changes to the previously reported information.

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 resulted in an increase of $2.16, and adding X-ALD to the newborn screening resulted in an increase of $10.84 per sample, for a total of $13 for both of these disorders. The $13 increase is included in the $138 screening fee that went into effect October 2019.

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

List all changes to the information reported on the Agency Background Document submitted for the previous stage regarding any requirement of the regulatory change which is more restrictive than

applicable federal requirements. If there are no changes to previously reported information, include a specific statement to that effect.

There are no changes to the previously reported information.

Agencies, Localities, and Other Entities Particularly Affected

List all changes to the information reported on the Agency Background Document submitted for the previous stage regarding any other state agencies, localities, or other entities that are particularly affected by the regulatory change. If there are no changes to previously reported information, include a specific statement to that effect.

There are no changes to the previously reported information.

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
Various	VDH received 64 comments from various members of the public in support of this regulatory action to add SMA and X-ALD to Virginia's newborn screening panel.	VDH concurs with the comments in support of the proposed regulatory amendment to add SMA and X-ALD to Virginia's newborn screening panel. No response is required.
Bret Rawlings, Virginia Hospital and Healthcare Association	Mr. Rawlings expressed concerns regarding the economic impact of the amended regulation on hospitals, birthing centers, midwives, and infants and their families. These concerns mainly address the costs highlighted in proposed stage agency background document and the Department of Planning and Budget's Economic Impact Analysis, which detail costs for the Department of General Services (DGS) Division of Consolidated Laboratory Services (DCLS) to implement the addition of SMA and X-ALD to the newborn screening panel. The costs are supported by fee increases to DCLS's dried blood spot specimen kits that went into effect in October 2019 and that these increased fees have been passed on to hospitals,	VDH concurs that this regulatory amendment and associated fee increases to DCLS's dried blood spot specimen kits potentially results in an economic impact to hospitals, birthing centers, midwives, and infants and their families.

	birthing centers, and infants and their families. Mr. Rawlings summarized that the approach to fund conditions that are added to the newborn screening panel by increasing fees for the dried blood spot kits is not sustainable. Mr. Rawlings recommended: "One possible option to address this in the Newborn Screening Regulations would be to limit DCLS authorization to establish fees or implement increases to fees to include only certain incremental variable costs incurred in performing tests. For example, the costs required to purchase additional instruments or equipment needed to perform the test, additional space and related build-out costs, and software costs could not be included in the fee. Such costs would either need to be absorbed by DCLS or funded through DCLS or Department appropriation requests to the General Assembly."	VDH concurs that there is an option to amend the regulatory language to limit fee increases related to adding new disorders to the newborn screening panel in the future. However, the fee increase to support the addition of SMA and X-ALD to the panel went into effect October 1, 2019. The regulation is currently undergoing periodic review and a stakeholder workgroup of the Newborn Screening Advisory Committee will review the regulation and make suggested amendments. The recommendation to amend 12-VAC5-71-100 will be put forth for consideration during the regulatory process. VDH does not concur that the costs to add new disorders to the newborn screening panel in the future should be absorbed by DCLS. VDH will follow the established process for developing and submitting a budget proposal request for a General Assembly appropriation for costs related to adding new disorders to the newborn screening panel in accordance with the regulation and when required. The costs for adding SMA and X-ALD to the newborn screening panel are supported by fee increases that went into effect October 1, 2019 so a request for an appropriation is not applicable at this time.

Detail of Changes Made Since the Previous Stage

*List all changes made to the text since the previous stage was published in the Virginia Register of Regulations and the rationale for the changes. 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. Explain the new requirements and what they mean rather than merely quoting the text of the regulation. * Put an asterisk next to any substantive changes.*

Current chapter-section number	New chapter-section number, if applicable	New requirement from previous stage	Updated new requirement since previous stage	Change, intent, rationale, and likely impact of updated requirements

		There are no new changes or requirements from the previous stage.		

Detail of All Changes Proposed in this Regulatory Action

*List all changes proposed in this action and the rationale for the changes. 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. Explain the new requirements and what they mean rather than merely quoting the text of the regulation. * Put an asterisk next to any substantive changes.*

Current chapter-section number	New chapter-section number, if applicable	Current requirements in VAC	Change, intent, rationale, and likely impact of updated requirements
12VAC5-71-30		This section lists the Virginia Newborn Screening System's core panel of heritable disorders and genetic diseases for which the newborn dried blood spot testing is conducted.	Change: 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.