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Notice of Intended Regulatory Action (NOIRA)
Agency Background Document

Agency name	Virginia Department of Health
Virginia Administrative Code (VAC) citation(s)	12VAC5-71
Regulation 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	March 5, 2019

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 (1 VAC 7-10), and the *Virginia Register Form, Style, and Procedure Manual for Publication of Virginia Regulations.*

Brief Summary

Please provide a brief summary (preferably no more than 2 or 3 paragraphs) of the subject matter, intent, and goals of this this regulatory change (i.e., new regulation, amendments to an existing regulation, or repeal of an 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 US Secretary of Health and Human Services' Advisory Committee on Heritable Disorders in Newborns and Children.

Acronyms and Definitions

Please define all acronyms or technical definitions used in the Agency Background Document. .

DCLS – Division of Consolidated Laboratory 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

Please 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, board decision, etc.). 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 Recommended Uniform Screening Panel (RUSP) of the US Secretary of Health and Human Services' Advisory Committee on Heritable Disorders in Newborns and Children.

Legal Basis

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

Please describe the specific reasons why the agency has determined that this regulation is essential to protect the health, safety, or welfare of citizens. In addition, please explain any potential issues that may need to be addressed as the regulation is developed.

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-linked adrenoleukodystrophy: 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 Virginia Department of Health's Office of Family Health Services has a longstanding partnership with DCLS to provide blood spot newborn screening services. The VNSP 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. The current newborn screening fee is $101.20 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. An estimated fee increase of $1.86 for SMA and an increase between $4.62 - $6.92 for X-ALD would need to occur at least twelve months prior to implementation to cover the cost of adding these screenings.

Substance

Please briefly identify and explain the new substantive provisions that are being considered, the substantive changes to existing sections that are being considered, or both.

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 Health and Human Services and the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children.

Alternatives

Please 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. 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.

Periodic Review and Small Business Impact Review Announcement

This NOIRA is not being used to announce a periodic review or a small business impact review.

Public Participation

Please indicate how the public should contact the agency to submit comments on this regulation, including ideas to assist the agency in the development of the regulation and the costs and benefits of the alternatives stated in this notice or other alternatives.

Also, indicate whether a public hearing is to be held to receive comments. Please include one of the following choices: 1) a panel will be appointed and the agency's contact if you're interested in serving on the panel is _____; 2) a panel will not be used; or 3) public comment is invited as to whether to use a panel to assist in the development of this regulation.

VDH is seeking comments on this regulation, including but not limited to: ideas to be considered in the development of this regulation, the costs and benefits of the alternatives stated in this background document or other alternatives, and the potential impacts of the regulation. The agency/board is also seeking information on impacts on small businesses as defined in § 2.2-4007.1 of the *Code of*

Virginia. Information may include: 1) projected reporting, recordkeeping, and other administrative costs; 2) the probable effect of the regulation on affected small businesses; and 3) the description of less intrusive or costly alternatives for achieving the purpose of the regulation.

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://www.townhall.virginia.gov. Written comments must include the name and address of the commenter. Comments may also be submitted by mail, email or fax to Robin Buskey, 109 Governor Street, Richmond, VA 23219, 804-864-7652, and robin.buskey@vdh.virginia.gov. 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 the proposed stage of this regulatory action.