Date of Hearing: April 23, 2024

ASSEMBLY COMMITTEE ON HEALTH
Mia Bonta, Chair
AB 2563 (Essayli) – As Introduced February 14, 2024

SUBJECT: Newborn screening program.

SUMMARY: Requires the State Department of Public Health (DPH) to expand statewide screening of newborns to include screening for Duchenne Muscular Dystrophy (DMD). Expands the purposes for which moneys from the Genetic Disease Testing Fund are expended.

EXISTING LAW:

1) Establishes the policy of the state to make every effort to detect, as early as possible, phenylketonuria (PKU) and other preventable heritable or congenital disorders leading to intellectual disability or physical defects. [Health and Safety Code (HSC) § 125000]

2) Requires DPH to establish a genetic disease unit to coordinate all programs in the area of genetic disease and to promote a statewide program of information, testing, and counseling services. Requires the unit to designate tests and regulations to be used in executing this program. [HSC § 125000]

3) Requires DPH to establish a program for the development, provision, and evaluation of genetic disease testing. [HSC § 125001]

4) Requires DPH to expand statewide screening of newborns to include tandem mass spectrometry screening for fatty acid oxidation, amino acid, organic acid disorders, and congenital adrenal hyperplasia as soon as possible. Requires DPH to provide information with respect to these disorders and available testing resources to all women receiving prenatal care and to all women admitted to a hospital for delivery. [HSC § 125001]

5) Requires DPH expand statewide screening of newborns to include screening for severe combined immunodeficiency (SCID) as soon as possible. In implementing the SCID screening test, requires DPH to also screen for other T-cell lymphopenias that are detectable as a result of screening for SCID, insofar as it does not require additional costs or equipment beyond that needed to test for SCID. [HSC § 125001]

6) Requires DPH to expand statewide screening of newborns to include screening for adrenoleukodystrophy (ALD) and any other disease that is detectable in blood samples as soon as practicable, but no later than two years after the disease is adopted by the federal Recommended Uniform Screening Panel (RUSP), or enrollment of the act amending this subdivision, whichever is later. [HSC § 125001]

7) Establishes the continuously appropriated Genetic Disease Testing Fund (GDTF), consisting of fees paid for newborn screening (NBS) tests, and states the intent of the Legislature that all costs of the genetic disease testing program be fully supported by fees paid for NBS tests, which are deposited in the GDTF. [HSC § 124977]

8) Authorizes moneys in the GDTF to be used for the expansion of the Genetic Disease Branch Screening Information System to include cystic fibrosis, biotinidase, SCID, and ALD and exempts the expansion of contracts for this purpose from certain provisions of the Public Contract Code, the Government Code, and the State Administrative Manual, as specified. [HSC § 124977]

FISCAL EFFECT: Unknown. This bill has not yet been analyzed by a fiscal committee.

COMMENTS:

1) **PURPOSE OF THIS BILL.** According to the author, this bill will add DMD to the statewide NBS panel. The author states that DMD is a debilitating and often fatal disorder that causes progressive muscle deterioration and weakness, affecting approximately one in 3,600 male infants worldwide. The author further notes that early intervention and treatment can make a critical difference in the life of a DMD patient, and further investment into understanding this disorder will bring us closer to more effective treatments and a cure. The author continues that this bill will bring California in line with the policies already enacted in states like Ohio and New York, along with many others currently considering similar proposals. The author concludes California can and should be a leader in this space, as NBSs for DMD will save and improve lives.

2) **BACKGROUND.**

   a) **DMD.** According to the Muscular Dystrophy Association, DMD is a genetic disorder characterized by progressive muscle degeneration and weakness due to the alterations of a protein called *dystrophin* that helps keep muscle cells intact. DMD symptom onset is in early childhood, usually between ages two and three. The disease primarily affects boys, but in rare cases it can affect girls. In Europe and North America, the prevalence of DMD is approximately six per 100,000 individuals. Muscle weakness is the principal symptom of DMD. It can begin as early as age two or three, first affecting the proximal muscles (those close to the core of the body) and later affecting the distal limb muscles (those close to the extremities). Usually, the lower external muscles are affected before the upper external muscles. The affected child might have difficulty jumping, running, and walking. Other symptoms include enlargement of the calves, a waddling gait, and lumbar lordosis (an inward curve of the spine). Later on, the heart and respiratory muscles are affected as well. Progressive weakness and scoliosis result in impaired pulmonary function, which can eventually cause acute respiratory failure. Life expectancy for those with DMD has increased over the years, with some patients surviving beyond 30 years. A 2015 study published in Pediatrics found that the prevalence of DMD has been estimated to be lower in Black individuals and higher in Latino individuals compared to white individuals. In terms of prevalence of DMD in California, there are five certified Duchenne Care Centers. The sponsor indicates these centers serve over 500 patients. It should be noted that this figure does not reflect all patients in California living with DMD, as there are patients who do not receive services from these centers.

   b) **Treatment of DMD.** In March 2024, U.S. Food and Drug Administration (FDA) approved Duvyzat (givinostat) oral medication for the treatment of DMD in patients six years of age and older. Duvyzat is the first nonsteroidal drug approved to treat patients with all genetic variants of DMD. It is a histone deacetylase (HDAC) inhibitor that works by targeting pathogenic processes to reduce inflammation and loss of muscle.

c) **Benefits of NBS.** NBS for DMD has been performed, primarily as pilot studies, in various parts of the world since the 1970s as early detection was believed to be beneficial. The benefits include early treatments such as physical therapy, allowing families to prepare for supporting a child with DMD by accessing appropriate resources and considering family planning options for future children. In 2023, Ohio became the first state to mandate universal NBS for DMD, with New York following shortly thereafter.

d) **Role of the federal government in NBS.** In the United States, screening of newborns is under the purview of state public health departments. Each state decides which disorders to screen, and expansion of each state's panel of screened conditions occurs on a state-by-state basis. The federal government also plays a role in NBS through the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) of the Secretary of the Department of Health and Human Services (DHHS). The ACHDNC is charged with evaluating conditions for screening newborns and children for heritable disorders recommending to the Secretary of DHHS the conditions for which newborns and children should be screened. If accepted by the Secretary, these conditions become part of the Secretary's RUSP which is made up of core and secondary conditions. Currently, there are 36 core and 25 secondary conditions on the Secretary's RUSP. ACHDNC also established a system of nomination and evidence review to evaluate conditions that are candidates for screening.

e) **California Newborn Screening Program (CNSP).** NBS began in California in 1966 with screening for one disorder, called PKU. The CNSP has expanded and now includes 80 different disorders, both genetic (passed down in families) and congenital (present at birth). To ensure the health of all its newborns, state law requires that all babies born in California have NBS soon after birth. The goal of the program is to identify babies with these disorders early, so that treatment can be started right away. A parent or guardian of the newborn child may only decline NBS based on the objection that it conflicts with his or her religious beliefs or practices. The CNSP takes a blood sample from a newborn's heel from 12 to 48 hours after birth to check for genetic disorders. At the same time, the newborn receives a hearing and congenital heart disease screening.

f) **Adding DMD to CNSP.** DMD screening is currently undergoing evidentiary review by ACHDNC. This bill would add DMD to California's Newborn Screening Panel. SB 1095 (Pan), Chapter 393, Statutes of 2016, requires DPH to expand statewide screening of newborns to include screening for any disease that is detectable in blood samples as soon as the disease as practicable, but no later than two years after the disease is adopted by the RUSP.

g) **Genetic disease testing.** In August 2023, the ACDHNC voted to move DMD screening to evidence-based review. The process is ongoing. For conditions that have been added to the RUSP using this process, the time from when a nomination is *first* presented to the Committee, to when the DHHS Secretary adds the condition to the RUSP has ranged from one year and nine months (21 months) to 10 years (120 months). Most have been around three to four years. According to information provided by the author's office, DMD screening may face an uphill battle for inclusion in RUSP because testing technology is nascent and evidence of success is limited. According to a review published in Nature in 2021, neonatal screening is considered for neonatal-onset disorders for

which early treatment shows strong evidence of improved outcome. The review further states that Although DMD does not fully meet these criteria, advocacy groups are in favour of an early diagnosis to enable early management and interventions. The review continues Emerging therapies that might be more effective when used in an early stage of the disease before irreversible muscle damage has occurred strengthen their request to include DMD in NBS.

3) **SUPPORT.** This bill is supported by various groups, including Cure Duchenne (CD)**,** who states that this bill would significantly reduce the diagnostic journey for families facing this devastating disease and enable them to access impactful treatments sooner. CCD continues that DMD is a devastating muscle disease. While rare, it is one of the most common forms of muscular dystrophy, occurring in approximately 1:5,000 male births. Those living with DMD lose their ability to walk, feed themselves, breathe independently and succumb to heart failure. CD further states that there is hope through new pharmacological and gene- based therapies, as there are now eight FDA approved treatments and more than 25 companies investing in finding a cure for this devastating disease. CD contends that while science is moving forward, diagnosis has not. Researchers and clinicians have been working for more than 20 years toward a genetic screening for DMD and yet the average age of diagnosis has remained at approximately five years old. This diagnostic delay is one too many families face—taking away precious time from patients and delaying access to the necessary medical guidance parents need to make informed decisions to give their child the best chance at a healthier, longer life. DMD can be detected at birth with the same drops of blood already used for the state's NBS panel. CD concludes that this bill will ensure California families will not be faced with the same diagnostic delays and give California children timely access to much needed care to provide their best chance at a healthier and longer life.

4) **PREVIOUS LEGISLATION.**

a) SB 643 (Pan) of 2017 would have added DMD to the list of medical conditions eligible for coverage under the Genetically Handicapped Persons Program. SB 643 was vetoed.

b) SB 1095 (Pan), Chapter 393, Statutes of 2016, requires the DPH to expand statewide screening of newborns to include screening for any disease that is detectable in blood samples as soon as the disease as practicable, but no later than two years after the disease is adopted by RUSP or enrollment of SB 1095, whichever is later.

c) AB 1559 (Pan), Chapter 565, Statutes of 2014, requires DPH to expand statewide screening of newborns to include screening for ALD as soon as ALD is adopted by RUSP.

d) AB 395 (Pan), Chapter 395, Statutes of 2011, requires DPH to expand statewide screening of newborns to include screening for SCID and, insofar as it does not require additional costs, other T-cell lymphopenias detectable as a result of screening for SCID, and would make related changes.

REGISTERED SUPPORT / OPPOSITION:

Support

Butte County SELPA
California Children's Hospital Assn
California Life Sciences
Capricor Therapeutics, INC.
CureDuchenne
National Association of Pediatric Nurse Practitioners (NAPNAP)
North Santa Cruz County SELPA
Satellos Bioscience INC.
SELPA Administrators of California
Solano County SELPA
Solid Biosciences
The Akari Foundation
Yolo County SELPA

Opposition

None on file.

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