Kennedy Krieger Institute

DATE: **February 3, 2022** **COMMITTEE: Senate Finance**
BILL NO: **Senate Bill 242**
BILL TITLE: **Maryland Department of Health - System for Newborn Screening - Requirements**
POSITION: **Support**

Kennedy Krieger Institute supports Senate Bill 242 - Maryland Department of Health - System for Newborn Screening - Requirements

Bill Summary:
Senate Bill 242 requires the Maryland Department of Health's Newborn Screening Program to screen for each condition listed in the U.S. Department of Health and Human Services' Recommended Uniform Screening Panel (RUSP).

Background:
Kennedy Krieger Institute is dedicated to improving the lives of children and young adults with developmental, behavioral, cognitive and physical challenges. Kennedy Krieger's services include inpatient, outpatient, school-based and community-based programs, which serve about 25,000 individuals every year. As part of these services, the Institute is involved in research and clinical care of individuals affected by rare genetic brain disorders. Over the last 5 decades many important research discoveries have led to establishing of new diagnostic tests and new therapies for many of these rare diseases.

Newborn Screening allows the early detection of treatable rare genetic disorders, resulting in a dramatic improvement in the lives of young babies and children, as well as their families (1). X-linked adrenoleukodystrophy (ALD) is a prototype example of rare genetic disorder that is currently identifiable through newborn screening (2). ALD is an inherited disorder of metabolism with recent data indicating an annual incidence of approximately 1 in 14,000 newborn males (3). It is a fatal disorder affecting the brain, the peripheral nervous system and the adrenal glands. Kennedy Krieger Institute, one of the first centers in the world to focus on ALD, currently provides care to about 500 patients affected by ALD. Internationally, Kennedy Krieger serves as both a major clinical and laboratory referral center. In fact, the newborn screening test that is being debated in SB242 was first developed at Kennedy Krieger (4) and has now been replicated by many laboratories across the United States and internationally.

Rationale:
Newborn boys with ALD appear perfectly normal and healthy at birth. They are discharged from the newborn nursery with their mothers and develop normally the first few years of life. About half of affected males will first develop behavioral problems around 5-7 years of life and then rapidly lose their vision, their hearing, and then their ability to talk, swallow, and to protect their airways. Next, boys with ALD become bedridden and either die or end up in a vegetative state, all within 2 years after symptom onset. Unfortunately, despite many attempts by various international groups, no effective therapy has been discovered for affected individuals once their first neurological symptoms have manifested (5).

Importantly, while diagnosis after symptom onset is too late for disease modifying therapies, the brain disease in ALD can be effectively halted by undergoing bone marrow transplantation, **but only if affected individuals are diagnosed prior to symptom onset** (6). Therefore, it is vital that affected individuals are diagnosed as early in life as possible to avoid severe suffering and death.

In 2014, following the death of their son, Aidan, owing to complications of ALD, the Seeger family drafted Aidan's Law which led eventually to establishment of ALD newborn screening in New York. In 2016, the then

US Secretary of Health and Human Services signed the recommendation to add ALD to the uniform panel of disorders screened in the newborn period in every state in the United States.

Since that recommendation from HHS in 2016, Kennedy Krieger has, on multiple occasions, petitioned the Maryland Department of Health and the State Newborn Screening Laboratory to establish ALD newborn screening in Maryland. In addition, we have offered to provide technical support for the laboratory testing, if necessary. In the interim, over 20 states in the United States have incorporated newborn screening for ALD into their panels. Unfortunately, Maryland has yet to adopt newborn screening for ALD.

In our opinion, it is extremely important that newborn screening for all condition listed under RUSP, including ALD, is implemented in the State of Maryland to save children affected by these rare disease from suffering and death.

Kennedy Krieger Institute requests a favorable report on Senate Bill 242.

References:

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2) Moser AB, Fatemi A. Newborn Screening and Emerging Therapies for X-Linked Adrenoleukodystrophy. JAMA Neurol. 2018 Oct 1;75(10):1175-1176.
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4) Theda C, Gibbons K, Defor TE, Donohue PK, Golden WC, Kline AD, Gulamali-Majid F, Panny SR, Hubbard WC, Jones RO, Liu AK, Moser AB, Raymond GV. Newborn screening for X-linked adrenoleukodystrophy: further evidence high throughput screening is feasible. Mol Genet Metab. 2014 Jan;111(1):55-7.
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6) Mallack EJ, Turk BR, Yan H, Price C, Demetres M, Moser AB, Becker C, Hollandsworth K, Adang L, Vanderver A, Van Haren K, Ruzhnikov M, Kurtzberg J, Maegawa G, Orchard PJ, Lund TC, Raymond GV, Regelmann M, Orsini JJ, Seeger E, Kemp S, Eichler F, Fatemi A. MRI surveillance of boys with X-linked adrenoleukodystrophy identified by newborn screening: Meta-analysis and consensus guidelines. J Inherit Metab Dis. 2021 May;44(3):728-739..