February 5, 2024

Dear Chair Pena-Melnyk

I write in opposition to HB96 (SB117). I strongly support the statement of the Maryland Advisory Council on Hereditary and Congenital Disorders (MACHCD).

I represent myself as a practicing Medical Geneticist and not any organization or institution.

To explain why my opinion is relevant to the legislators charged with the welfare of the children of Maryland: I have served as the lead clinical Geneticist for the University of Maryland School of Medicine since 2004. I diagnose and treat genetic conditions. I see the impact of the late diagnosis of treatable genetic conditions on children and families. It is my passion to identify those with treatable conditions as early as possible.

To that end, since the 1980s in Colorado I have been actively involved in the responsible expansion of newborn screening panels. Beginning in 1999 I was involved at the national level, as a member of the staff for a U.S. Senate committee working to introduce the federal bill that led to creation of the federal Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC). I then worked for four years for the U.S. Department of Health and Human Services, including some collaboration with the ACHDNC. Since my return to academic clinical practice, I continued to work on responsible expansion of newborn screening as an officer of the Society for Inherited Metabolic Disorders (SIMD), the professional organization for professions treating and studying inborn errors, including as the SIMD's liaison to the ACHDNC.

**I very strongly support the process by which the United States and the State of Maryland currently evaluate and make recommendations for the expansion of newborn screening.**

A major goal of the federal ACHDNC is to assure all babies are tested for treatable conditions, regardless of the State of residence. To do that, the ACHDNC created the Recommended Uniform Screening Panel (RUSP). The ACHDNC invests the dollars needed for rigorous evaluation of each candidate condition. That evaluation is then considered by ACHDNC committee members who are parents/advocates, scientists, clinicians and public health experts. These stakeholders make the decision about nomination to the RUSP.

The State of Maryland legislature has wisely taken advantage of the Federal resource while maintaining the ability of the State to chart its own course. By statute, Maryland's Advisory Committee (the MACHCD) includes parents and other advocates as well as clinical and public health experts – and two members of the Maryland legislature. Since its inception, the MACHCD makes independent recommendations to the Maryland Secretary of Health for addition to Maryland NBS panel, and has open meetings and invites public input. More recently the law was amended to require the MACHCD to respond to any Federal addition to the RUSP. The MACHCD does all of these things in a responsible manner for the good of babies and families in Maryland.

As a physician, I appreciate the long-standing support of the Maryland government for newborn screening and for the follow up diagnosis and treatment of babies with genetic conditions. I see the effects of newborn screening on babies and families. I partner with families from the moment we tell them that newborn screening alerts us that a baby might have a serious condition. My team and I work with the family to test the baby as fast as possible. For each

affected baby, my team treats inborn errors of metabolism, and together with families we celebrate the success of treatment.

I also see the effects on babies and families when newborn screening tells us a baby might have a genetic condition and none is found – the "false positive" screens. False positives are not lab errors. They are a result of the physiologic overlap of the levels of the markers of the condition in the blood and therefore they are unavoidable.

NBS also has risk. When NBS identifies a baby at risk for a serious condition, families become anxious, sometimes even terrified. We work to allay that anxiety. I have worked with the support group Genetic Alliance's newborn screening arm Baby's First Test to study this issue. Together we developed and disseminated strategies to support families though the journey when NBS finds a baby to be at risk. However, when NBS leads to a need for invasive testing, I cannot eliminate the risk of injury to the baby from the testing.

In medicine we consider a blood test "invasive" but it is a minor procedure, with little risk of injury. But even a simple blood draw on a newborn is traumatic for many families. I know this because of what the families say, and because I am often the one drawing the blood sample, with parents sometimes crying and needing to leave the room. And that is just a blood sample.

For Krabbe, the current follow up after alert from NBS involves much more than a blood test, it requires MRI scan and spinal tap. An MRI is not usually dangerous for a newborn since we typically do not need to sedate a newborn for the test. But for the infant, toddler or older child who cannot hold still in the MRI machine, sedation with its attendant risks is necessary. Each MRI means bringing the child into the medical setting, with risk of exposure to infections and possible high cost to the family. And a spinal tap - inserting a needle between the vertebrae to collect cerebral spinal fluid - is a procedure with more risks.

If Krabbe had been added in the past to the Maryland NBS panel, every baby with a positive screen would have needed an MRI and spinal tap to find out whether the baby 1) had infantile Krabbe and needed bone marrow transplant, 2) was at risk for late onset Krabbe, or 3) did not have Krabbe at all. Babies at risk for late onset Krabbe then need repeated MRIs and spinal taps for years. And in New York, for every baby found with infantile Krabbe, there were four babies found at risk for late onset Krabbe. Recent scientific advances are improving the screening for infantile Krabbe, and I expect it will become appropriate to add it to the newborn screening panel – indeed that it is right now in process of addition to the RUSP. But I think that is a decision that should be made by the existing thoughtful and responsible process and not by legislation.

I know that this Committee and all Maryland legislators want the best for all Maryland children and families. However, **the proposed bill would set a dangerous precedent that interferes with a complex process wisely set up by this legislature and that currently works well. I strongly urge you to continue to trust the process currently in place in accord with existing Maryland law.** I would be happy to answer any questions.

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