



# University of Pittsburgh

## *School of Medicine*

Children's Hospital of Pittsburgh  
Division of Medical Genetics  
4401 Penn Avenue  
Pittsburgh, PA 15224  
(412) 692-7746  
Fax: (412) 692-7816  
[gerard.vockley@chp.edu](mailto:gerard.vockley@chp.edu)

**Jerry Vockley, M.D., Ph.D.**  
**University of Pittsburgh**  
*Cleveland Family Professor of Pediatric Research*  
*Professor of Human Genetics*  
**Children's Hospital of Pittsburgh of UPMC**  
*Chief of Medical Genetics*  
*Director of the Center for Rare Disease Therapy*

April 16, 2017

Dear Representative Cruz and Committee Members:

Thank you for the opportunity to provide written testimony on the topic of newborn screening in the Commonwealth of Pennsylvania. I regret that I am out of the country and unable to appear before the Committee in person. First, let me introduce myself and my experience regarding newborn screening. I am Professor of Pediatrics and Human Genetics at the University of Pittsburgh Schools of Medicine and Public Health, and Chief of Medical Genetics and Director for the Center for Rare Disease Therapy at the Children's Hospital of Pittsburgh of UPMC. Born and raised in Pittsburgh, I received my MD and PhD at the University of Pennsylvania, trained in Pediatrics and Human Genetics and became Chair of the Department of Medical Genetics at the Mayo Clinic before returning to Pittsburgh in my current capacities in 2004. I have served on the Minnesota Newborn Screening Advisory Committee, and, for the last 10 years on the Pennsylvania Newborn Screening Technical Advisory Board (the last several as its Chair), and previously served on the HRSA Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC), tasked with developing the Recommended Universal Screening Panel (RUSP). I have spent all of my professional career advocating for babies and parents, and view newborn screening as one of the most important public health initiatives undertaken by our Department of Health.

The PA Newborn Screening Technical Advisory Board (TAB) was constituted by law in 1980 to provide advice to the Secretary of Health on matters of newborn screening. It neither makes policy, nor dictates the diseases mandated for newborn screening in Pennsylvania. Following careful review of the medical evidence, the TAB makes recommendations to the Secretary, who is free to accept or reject its advice.

Newborn screening has been designated by the Centers for Disease Control and Prevention as one of the 10 most successful public health programs of the 20<sup>th</sup> Century. In large part, its success has been predicated by careful application of the tenets of public health policy. Screening for a disease in newborns is only viewed as appropriate if the disease meets very specific criteria:

- It has significant impact in babies
- A sensitive and specific screening test is available
- Early treatment of the disease affects its outcome

- Sufficient monetary resources are made available to not only pay for screening, but for follow up and support of the infants identified as affected

Addition of new diseases to the newborn screening program that do not meet these requirements by definition leads to inappropriate use of limited resources. In Pennsylvania, newborn screening was limited to six diseases mandated in 1980. Passage of Act 36 in 2008 created two lists of diseases, those mandated by the State for screening and those mandated for follow up by the State if screening was performed. At that time, the original six diseases were added to the former list, and all others were added to the latter as they were already being screened voluntarily by hospitals. Since then, the TAB has consistently recommended the elimination of the mandated follow up list and transfer of all diseases to the mandated screening list. Limitations in Department of Health budget have kept both lists intact, as the Department of Health has indicated that it must pay laboratory costs for conditions on the mandated screening list but not conditions on the mandated follow up list from its existing budget.

Much controversy has arisen regarding addition of six new disorders to newborn screening in 2014 through Act 148. These six diseases are all rare genetic disorders known as lysosomal storage diseases and include:

- Krabbe disease
- Gaucher disease
- Nieman-Pick disease, type A and B
- Hurler syndrome (MPS1)
- Pompe disease
- Fabry disease

The TAB agreed that MPS1 and Pompe disease, recently added to the RUSP by the HRSA Secretary's ACHDNC, should be incorporated immediately into mandated PA screening, but felt that inadequate evidence existed for addition of the others (which have not been added to the RUSP) to the mandated screening program. Nieman-Pick disease is not treatable, while Gaucher and Fabry diseases rarely present clinical symptoms in babies or young children. While they can be added to the State screen for minimal additional cost for the initial screening test, complicated and expensive testing to confirm the diagnosis ultimately leads to a cost of approximately \$750,000 for each disease added. Given that no additional appropriation accompanied Act 148, the TAB did not find it justifiable to recommend they be added to the mandated screening panel and instead suggested temporary addition to the mandated follow up panel, whose costs are covered by medical insurance and/or hospitals, pending identification of additional resources to cover expenses related to further expansion of the panel.

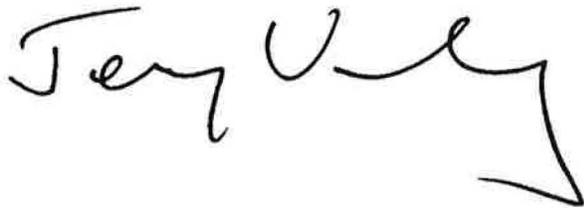
Krabbe disease has been more controversial because of legitimate differences of opinion in the medical field. Most newborn screening experts, including the HRSA panel have deemed medical evidence of efficacy of treatment of babies identified by newborn screening as inadequate. In addition, many babies identified in a pilot program in New York State were identified as questionably affected and underwent an invasive and dangerous series of medical tests before ultimately being declared unaffected. Metabolic physicians in New York, along with the American College of Medical Genetics, ultimately declared the pilot program a failure and recommended against further screening for Krabbe disease. Recent advancements in the screening algorithm and early treatment are promising but have not yet been published in a peer reviewed journal. Thus, while the TAB remains committed to reassessing new information

as it comes forward, there is still insufficient medical evidence to yet change its previous recommendation on this disease.

The Legislature is now considering House Bill 1081 to revamp the newborn screening infrastructure in PA. There is much to like about this bill. As Chair of the TAB, I applaud and fully support the institution of a screening fee to help fund the program outside the usually budgetary process. Such a fee will allow flexibility in expanding newborn screening and provide sustainable resources to pay for appropriate screening, follow up, and treatment of infants for diseases on the mandated screening list. I also support the establishment of a single mandated screening list. I agree that addition to or subtracti from the list should only be on the basis of TAB review of the medical evidence and acceptance of a recommendation to add by the Secretary of Health. The bill includes a long list of diseases to be added to the mandated screening risk. I respectfully suggest that inclusion of specific disorders in the bill will make it more difficult for the Secretary of Health to manage the mandated screening list and remove disorders that might ultimately be shown not to be appropriate for screening. In general, the PA newborn screening panel should follow the RUSP with only rare exceptions for diseases and unique circumstances that call for addition to the PA panel outside the RUSP.

I greatly appreciate the Legislature's interest in the health of the babies of Pennsylvania in general and newborn screening in particular, and look forward to working with you on advocating for their best care.

Regards,

A handwritten signature in black ink, appearing to read "Jerry Vockley". The signature is fluid and cursive, with a long, sweeping tail that extends downwards and to the right.

Jerry Vockley, M.D., Ph.D.