

**SHELLY HETTLEMAN**  
*Legislative District 11*  
Baltimore County

Chair, Rules Committee  
Budget and Taxation Committee

*Subcommittees*  
Capital Budget  
Health and Human Services  
Chair, Pensions

*Joint Committees*  
Senate Chair, Audit and Evaluation  
Senate Chair, Pensions



**THE SENATE OF MARYLAND**  
ANNAPOLIS, MARYLAND 21401

James Senate Office Building  
11 Bladen Street, Room 220  
Annapolis, Maryland 21401  
410-841-3131  
800-492-7122 Ext. 3131  
Shelly.Hettleman@senate.state.md.us

**TESTIMONY OF SENATOR SHELLY HETTLEMAN**  
**SB 495 - HEALTH - NEWBORN SCREENING PROGRAM - GAUCHER**  
**DISEASE**

Madam Chair, Mr. Vice Chair, and members of the Senate Finance Committee, I am writing to express my strong support for **SB 495 - Health - Newborn Screening Program - Gaucher Disease**.

**What Is Gaucher Disease?**

Gaucher Disease is a rare genetic condition that causes a buildup of fatty deposits throughout the body, which can lead to progressive organ damage, chronic pain, anemia, bone disease, and, in severe cases, life-threatening complications. Although Gaucher Disease affects individuals across all racial and ethnic backgrounds, it disproportionately impacts people of Ashkenazi Jewish descent. The incidence of Gaucher Disease in this population is approximately 1 in 450 individuals, compared to roughly 1 in 100,000 in the general population.<sup>1</sup> This disparity underscores the importance of equitable public health measures that ensure early identification and intervention for communities at elevated genetic risk, while also recognizing that Gaucher Disease can affect any child born in Maryland.

Regardless of background, without early diagnosis, the symptoms of Gaucher Disease may go unrecognized for years, during which time irreversible harm can occur. Newborn screening for Gaucher Disease is both feasible and effective. Reliable screening methods already exist and are used in several jurisdictions worldwide, enabling early identification and timely intervention.

Maryland law mandates that the Maryland Department of Health screen for all disorders on the Recommended Uniform Screening Panel (RUSP), which provides federal recommendations for disorders to be included on state newborn screening panels. Each year, newborn screening performed per the RUSP identifies approximately 14,000 infants with serious conditions that benefit from early intervention.<sup>2</sup> However, despite evidence that newborn screening enables timely monitoring and treatment to prevent irreversible organ damage and reduce disease complications, Gaucher Disease is not currently part of the RUSP. SB 495 fills this gap.

## **Federal Changes and Their Impact on Maryland**

Historically, Maryland relied on vetting from the federal Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) to add new disorders to its newborn screening panels. The role of the ACHDNC was to evaluate scientific evidence and make recommendations to the U.S. Secretary of Health and Human Services regarding which conditions should be added to the RUSP. However, the ACHDNC was dissolved in April 2025, creating uncertainty about the future of disorders being added to the RUSP.

Following the dissolution of the ACHDNC, Maryland's State Advisory Council on Hereditary and Congenital Disorders (the Council) established a transparent and objective process for nominating, reviewing, and recommending new disorders to add to the Maryland newborn screening panel. The Council comprises experts in genetic disorders and community members with lived experience who review scientific literature to determine if a disorder meets inclusion criteria for newborn screening. These criteria include the availability of a reliable test, effective treatment, and evidence that the disorder causes irreversible harm to the child during the newborn period if left untreated. In the absence of federal RUSP inclusion, a disorder can now be added to the Maryland newborn screening panel through this new Council review and recommendation process.

## **The Role of SB 495**

Maryland has already taken a proactive and thoughtful step by empowering its State Advisory Council on Hereditary and Congenital Disorders to evaluate disorders using transparent, evidence-based criteria. This process ensures that decisions about newborn screening are grounded in scientific rigor, clinical effectiveness, and ethical responsibility. Gaucher Disease is precisely the type of condition this process was designed to evaluate.

Yet, this process is still evolving and does not guarantee timely implementation. SB 495 directly addresses this gap by requiring the Maryland Department of Health to include Gaucher Disease in the state's newborn screening program without unnecessary delay. This ensures that infants in Maryland are not left vulnerable to preventable harm due to federal delays in RUSP inclusion.

Newborn screening is one of the most effective public health interventions available. It is designed to identify serious conditions before symptoms appear, when intervention can make the greatest difference. By adding Gaucher Disease to Maryland's newborn screening program, SB 495 affirms the state's commitment to preventing avoidable harm, promoting health equity, and using science-based evidence to guide our policy.

Therefore, I respectfully urge a favorable report on SB 495.