My name is Shanna Quimby and I am testifying in memory of my son Gavin Quimby. Gavin was diagnosed with Metachromic Leukodystrophy(MLD) when he was three in December of 2013 and died from complications of this disease in September of 2015 at the age of five. Gavin was a funny, sweet, and empathetic kid, his personality and smile won over everyone's heart who was in his life. He was our beautiful boy who was taken too soon. MLD is a swift and devastating disease that is difficult to be successfully treated without early detection. Gavin's diagnosis came too late and the journey there was long and painful. Every minute counts with this disease, the sooner the child is detected with MLD, like at birth, the sooner treatment can begin. Also, Minnesota has a world renowned Leukodystrophy center at The University of Minnesota's Masonic Children's Hospital, these babies would have the best chance in the world to be treated successfully and live the life that Gavin couldn't. If this bill passes there will hopefully never be another Minnesota parent who will watch their child die from this terrible disease.

Written Testimony from Prof Michael Gelb, Univ of Washington, Dept. of Chemistry

My lab at the Univ. of Washington developed the method for newborn screening of MLD. We carried out the first pilot study by testing 30,000 newborn dried blood spots using the method (analysis of the sulfatide lipid and then the ARSA enzyme, both by mass spectrometry). We found 1 true case of MLD and zero false positives (which is a remarkable result). As far as false negatives, we expect none based on the fact that when we took 40 newborn dried blood spots from patients that went on to develop and be diagnosed with MLD, and all of them tested positive.

The mass spectrometry newborn screening assay for MLD can be added to ongoing panels of diseases now being screened across the USA including the Minnesota newborn screening lab. Laboratories will need to add a liquid chromatography module to their existing mass spectrometry equipment, which increases the equipment cost by about 20% (some labs have this already). Once the module is added, the following diseases can be done in a single analysis at ~2 min per newborn: 1) MPS-I and Pompe disease (now carried out in the Minnesota newborn screening program); 2) MPS-II and Krabbe disease (being added soon to the Minnesota program as they are now on the Recommended Screening Panel); 3)X-linked adrenoleukodystrophy (now on the Minnesotta panel or being added soon); 4) MLD. This analysis is easily expandable to include other diseases that are likely to be added to the state newborn screening panels in the coming few years. All reagents to add MLD are commercially available including assay reagents and validation standards. Initially labs can add MLD as a laboratory developed test, and an FDA-approved MLD newborn screening method is expected in 2026.

MGELB