I developed the first method used in the world for newborn screening of Krabbe disease. It is used in the following newborn screening labs in the USA:

NY, IL, KY , OH, and TN.

It is based on mass spectrometry, and an FDA-approved kit is available from PerkinElmer Corp.

The only other state doing newborn screening for Krabbe disease is MO, and they use a fluorescence method, not FDA approved, but a laboratory developed test.

It is my understanding that OR now screens for 4 lysosomal storage diseases: Pompe, MPS-I, Gaucher, and Fabry, using a fluorescence assay based on digital microfluidics sold by Baebies Corp. Most other states doing newborn screening for lysosomal storage diseases use mass spectrometer. If OR would switch to my mass spectrometry method, they could do these 4 diseases plus Krabbe disease in the same multiplex analysis. There is no digital microfluidics fluorescence assay for Krabbe disease. MO carries out a fluorescence assay for Krabbe disease using a standard plate-reader fluorimeter. If OR were to adopt this method, it would cost about $0.2-0.3 per newborn for reagents, and would require a part time lab tech and a fluorescence plate reader that cost about $20,000. There are additional costs of newborn screening, but these are good estimates for the indicated items.

Based on more than a decade of experience for newborn screening for Krabbe disease outside of OR, we have learned how to well classify newborns into 3 categories: 1) have infantile Krabbe disease with certainty, 2) are at high risk to develop a later onset form of Krabbe disease in early childhood, 3) no risk to develop Krabbe disease. Based on the numbers of patients in these categories in NY we can predict with high confidence that OR will find 0-1 category 1 and 0-1 category 2 patients per year (simple population scaling). Category 1 newborns are referred immediately to treatment centers, whereas category 2 newborns are recommended for followup medical examinations on the order of every few weeks for the first few months of life and then less often until a few years of age. Parents would be advised about the type of symptoms that may turn up. Although this creates family anxiety, the numbers are small (as stated about 0-1 newborns pear yr). In summary, after more than a decade of experience, we know how to screen for Krabbe disease with high certainty.

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