

Illinois Department of Public Health  
Lysosomal Storage Disorders Subcommittee  
Illinois Department of Public Health  
Meeting and Conference Call Minutes: December 2, 2015

Subcommittee Members Attending

Kelie Bontempo, Shantel Buhl, Advocate Lutheran General Children's Hospital  
Barbara Burton, Chair, Stacy Bichl, Katherine Keating, Lauren Whitaker, Lurie Children's Hospital  
Jennifer Burton, University of Illinois College of Medicine at Peoria  
David Dimmock, Medical College of Wisconsin  
George Hoganson, Rich Dineen, University of Illinois Chicago  
Linda Manwaring, St. Louis Children's Hospital  
Tess Rhodes, DSCC  
Natasha Spencer, Consumer Representative  
Darrel Waggoner, Maria Helgeson, University of Chicago

IDPH Staff

Rebecca Barnett, Khaja Basheeruddin, Jean Becker, Matt Charles, David Culp, Shannon Harrison, Tom Johnson, Claudia Nash, Conny Moody, Rong Shao, Heather Shryock

The meeting was called to order at 4:04 p.m. Dr. Barbara Burton introduced the consumer representative, Natasha Spencer. At this time, Ms. Spencer read a brief story of her son's struggle with Krabbe disease (attached). She questioned the time taken to find a solution for Krabbe molecular testing as discussed on the last call.

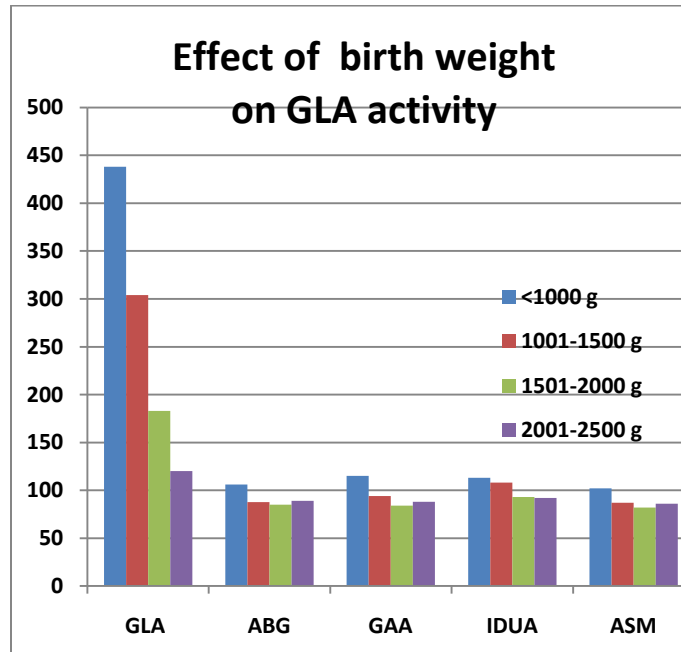
IDPH Laboratory Status Report

Khaja Basheeruddin provided LSD enzyme activities expressed as percent of daily median for different birth weight groups. He analyzed data of 60,000 – 70,000 samples and divided them into groups based on birth weight, and noted GLA was the only one behaving differently than other four enzymes. Based on this, the lab made a recommendation:

- Lift cut-off for GLA to 50% of the daily median for specimens having <2000 g birth weight
- Specimens having GLA activity <50% will have LSD result code as "Invalid", requiring a resubmitted specimen.
- The lab will request a second specimen after 3 weeks (expected resubmission rate = 2-3 samples/week)

	<b>&lt;1000 g</b>	<b>1001-1500 g</b>	<b>1501-2000 g</b>	<b>2001-2500 g</b>
<b>(n)</b>	<b>412</b>	<b>524</b>	<b>1159</b>	<b>3644</b>
<b>GLA</b>	438	304	183	120
<b>ABG</b>	106	88	85	89
<b>GAA</b>	115	94	84	88
<b>IDUA</b>	113	108	93	92
<b>ASM</b>	102	87	82	86

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After much discussion, the recommendation was **not** passed at this time. Dr. Burton suggested reporting as invalid if less than three or four weeks of age and work on developing language for reporting low birth weight infants. The committee would like follow-up data as quickly as possible with written documentation for the detection of all five diseases showing pending, normal, unresolved.

#### Consented Case Discussion

At this time, the specialists and genetic counselors discussed specific cases by institution. IDPH staff indicated they would be sending out notification to genetic counselors that the cutoff date for submission of data will be one week prior to scheduled subcommittee calls, and all consent forms and definitive diagnosis forms must be submitted to IDPH to be included on the spreadsheet of cases to be reviewed.

#### Open Discussion

Dr. Hoganson again brought up the discussion about lack of available resources to pay for diagnostic testing. He asked if there could be anything done at the legislative level since it is a mandated test. Deputy Director Moody indicated that health insurance plans were not required to cover all diagnostics, so therefore there was no state legislation mandating coverage of diagnostic testing. There was a possibility of finding a legislator to prepare legislation to cover certain testing. Conny said she would look into additional information at the Department of Insurance. She asked Dr. Burton for to prepare the language for third party payor coverage to provide the Department of Insurance.

Dr. Dimmock requested DSCC qualifying forms. Tess Rhodes from DSCC will send the forms to Dr. Dimmock.

The next conference call is scheduled for **December 2<sup>nd</sup> at 4:00 p.m.** The meeting adjourned at 5:03 p.m.

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**Statement from Consumer Representative Natasha Spencer given on the 9/23/15 LSD Call:**

*This is in regard to screening for Krabbe:*

*With all due respect to everyone here who has worked hard and has endured the ups-and-downs getting this program running (I am aware of the timeline through updates from Claudia Nash), YOU ARE TAKING TOO LONG. It shouldn't take eight years. On behalf of two other IL families and my own, I sent a letter to Dr. Shah saying so. I am not a parent who's out there advocating for NBS like a dog with a bone. I personally struggle on a daily basis with what's right. Krabbe is a wretched disease and a transplant has numerous variables that are beyond our control. I experience conflict having given my son a G-tube. By doing so, I've asked him to live through much more than the disease itself would. But, YOU ARE STILL TAKING TOO LONG.*

*Kenan, who was born in 2011, has 2 copies of the 30kb Deletion, the most aggressive form of the disease, but is presenting as Late Infantile. He is now 4.5 years old and is aware of his environment. He can throw me a look when I walk into the room that says, Mom, suction me because the nurse isn't doing a good enough job. And if he can do that than he is also aware of how difficult it is to breath. His biggest issue currently is the effect of his low tone on respiration. Open your mouth as wide as you can without straining and inhale. You are breathing with your throat muscles.*

*Kenan was not transplanted and is living through this disease anyway.*

*Early detection is valuable for other reasons. There is an infant from KT born in December 2014, who was trached for choking on his secretions. A trach! There are suctioning techniques for that. And now complications and more surgery! As a parent, you are at the mercy of your doctors. The family was not put in touch with Hunters Hope, or our FaceBook Krabbe Community or referred to Dr. Maria Escolar, who now has a Mobile Medicine Unit and is seeing patients in clinic remotely eliminating the cost and inconvenience of traveling to Pittsburgh.*

*When I hear there is turn over in staff at University of Illinois Chicago Medical Center delaying this another 6 months, THAT is not justification for another child and family to live through a variation of our scenario. I don't think I need to convince any of you of this, you are all on the front lines in one way or another, but perhaps I can ask you to reinvest yourself in a sense of urgency. YOU ARE TAKING TOO LONG!*