

# **State Advisory Council on Hereditary and Congenital Disorders**

**Minutes February 12, 2014**

## **Members Present**

Miriam Blitzer, PhD, Acting Chair  
Erin Strovel, PhD, (nominee to Council)  
Hilary Vernon, MD, (nominee to Council)  
Caryl Siems  
Neal Porter, MD  
Anne Eder  
Sandra Takai, MD

## **Ex-Officio Present**

Fizza Majid, PhD  
Deborah Badawi, MD  
Robert Myers, PhD  
Lee Woods, MD

## **Members Absent**

Delegate Shirley Nathan-Pulliam  
Coleen Giofredda  
Anika Wilkerson

## **Staff**

Johnna Watson, RN (scribe)  
Linda Lammeree, RN  
Angela Sitler  
Tina Wiegand

## **Guests**

Carol Greene, MD  
Katie Bisordi  
Douglas Mogul, MD  
Tiffany McNair, MD  
Julie Hoover-Fong, MD

**Called to Order – 6:00 pm**

### **I. Welcome and Introductions**

Members and attendees introduced themselves.

### **II. Approval of October 2013 Minutes**

Minutes reviewed and Dr. Greene requested addition to the minutes to include comments made by herself and Dr. Hamosh regarding the quality of the currently published data regarding treatment for lysosomal storage disorders. Notes were given to Dr. Badawi regarding these comments.

### **III. Special Discussion**

- **Newborn Screening for Biliary Atresia**
  - Presentation given by Dr. Mogul, pediatric gastroenterologist at Johns Hopkins.
  - Biliary atresia is most common cause of liver transplants in US.
    - ✓ Occurs in 1:8,000-18,000 births (higher prevalence seen in Asian population)
    - ✓ In US, average age to diagnosis is 70 days. Outcomes would be improved with shorter time to diagnosis
    - ✓ Approximately 5-8 cases/year is predicted rate in Maryland
  - In 2004, Taiwan adopted stool color card screening
    - ✓ Pale stool is indicative of biliary atresia
    - ✓ Sensitivity is 97% and specificity is 99.5%
    - ✓ 5 year survival increased from 56 to 89%
    - ✓ Taiwan has a registry to track screening (80% response rate)

- Potential Screens are in development
  - ✓ Measurement of direct bili. In biliary atresia, direct bili increases in first 24 hours.
  - ✓ Measurement of bile acids in dried blood spots. Taurocholate (a bile acid) performed best with sensitivity of 79%
  - ✓ Mobile Health application for smartphones – plan is to use interactive color matching software to identify acholic (lack of bile) stools. The app would remind parents to check the color of their baby's stool at 2, 4, 6 and 8 weeks of age. If the stool is too pale, there would be an automatic reminder to call the baby's primary care provider.
- There is a current small pilot study in Hopkins nursery.
  - ✓ The baseline knowledge regarding stool color is being assessed, along with anxiety of the mother. Education given and then knowledge and anxiety are reassessed.
- Dr. Mogul states none of the states has done a large group study at this time. He wants to explore possibility of conducting one in Maryland.
  - ✓ Dr. Greene commented that by calling it a NBS DHMH would have to be involved, and Dr. Badawi indicated it should be a standard of care through AAP. Dr. Mogul said he is trying to get the screen placed on the neonatal periodicity table but wants to conduct a large scale study to prove effectiveness.
  - ✓ Dr. Badawi explained that all hospital births are currently in a database and DHMH might be able to support a compliance rate study.
  - ✓ Dr. Blitzer suggested starting with a few of the large birth hospitals in the State.
  - ✓ Dr. Mogul is exploring funding opportunities at this time and is in discussion with Proctor and Gamble currently to possibly add something to their diaper boxes.
  - ✓ Dr. Greene also offered that Joint Commission is trying to get total bili done on every baby at 24 hours of age so may be able to add direct bili as well.
- Dr. Mogul will continue to talk with Dr. Badawi regarding this issue.

#### **IV. Old Business**

- **Newborn Screening for Lysosomal Storage Disorders**
  - Dr. Blitzer reported that SB 433/HB891 also known as Lily's Law was introduced in this legislative session.
    - ✓ Hearing held by Finance Committee last week.
    - ✓ Laboratories Administration put in a fiscal note.
    - ✓ DHMH opposed bill secondary to process of review – The addition of disorders to the newborn screening panel is not legislated, but done through review process by the Council. Dr. Badawi indicated it is difficult to guess if the bill will go out of committee although some legislators seemed to be swayed by the family's testimony.
    - ✓ Dr. Greene stated that Krabbe disease was rejected for addition to the RUSP by the Federal Advisory Council prior to formal review.
    - ✓ NY currently screens for Krabbe disease. Missouri is performing a statewide pilot screening study. A few other states have passed legislation but are not screening yet..
    - ✓ Dr. Majid stated there is no FDA approved test for Krabbe disease screening at this time.
    - ✓ Hearing is not scheduled currently in the House.
    - ✓ Dr. Greene indicated that if screening is done using DNA will also identify carrier status and this will need to be addressed as well.
- **Update from NBS Lab/Laboratories Administration**
  - Dr. Myers reported that he has asked for supplemental funding for SCID NBS in the Governor's budget. No response at this time.
    - ✓ Anne Eder stated that March of Dimes has an Advocacy Day in the legislature and will include funding for SCID NBS in their plans. Will look into how to expand into meeting with the Governor's Office. Dr. Badawi stated that legislators do have some pull in adding to the supplemental budget.
    - ✓ Payments of the NBS fee, which is currently \$100, goes into general fund and then may be reallocated to the Lab, but the funds do not go directly to support the NBS lab.
  - Move to new lab will be started in late May or early June.

- Dr. Majid reported that they have been working with submitters to increase timeliness of specimen submission. Goal is to have 90% of specimens received with 72 hours after collection. Measuring from 10/1-12/31/2013, 19 hospitals are above 90% now. Graphs are given to the hospitals for comparison. Dr. Greene suggested the graphs also go to Risk Management and Caryl Siems also suggested Patient Safety Committee.
- **Update from OGSPSHCN**
  - Dr. Badawi reported that Maternal and Child Health Grant has been level funded which will result in some cuts that have been delayed from previous years.
- **Update on NCAA and sickle trait pilot project**
  - Dr. Blitzer reported that University System of MD is proposing to develop an educational program for the different campuses about sickle trait screening in athletes. Dr. Greene said that Alexia Thomas with HRSA is head of a program there discussing the NCAA screening policy and impact on NBS labs. Presentation on this program will be given at the next meeting.

## **V. New Business**

- **Membership Changes and Election of New Chair**
  - There are 11 voting representatives on the Council
    - ✓ There is 1 non-health related vacancy
    - ✓ There is also a vacancy from the Senate
  - Dr. Blitzer moved that Anne Eder be elected Chair of the Advisory Council
    - ✓ The nomination was seconded and voting members elected Anne Eder as Chair
- **Current Legislation in General Assembly**
  - Dr. Badawi updated the Council on the Senate hearing on SB 433 (adding lysosomal storage disorders to newborn screening panel)
    - ✓ Mr. and Mrs. Smith (Lily's parents) gave testimony, along with her grandmother.
    - ✓ Difficult to determine how committee members will vote
    - ✓ DHMH does not support the bill on the basis that addition of disorders to the newborn screening panel should be determined by scientific review and not by legislation.
    - ✓ Subcommittee of this Council appointed to review lysosomal disorders for possible future addition to the newborn screening panel.
  - HB 906 – Direct to consumer genetic testing
    - ✓ Laura Herrera submitted a report which indicated DHMH thinks this type of testing may not be a good idea. DHMH support is not known at this time
    - ✓ Dr. Greene suggested there should be a disclaimer that an individual has to talk to a healthcare provider prior to testing.
  - HB 761 – Specialty Drug Coverage
    - ✓ limits copays for specialty drug coverage
    - ✓ No position from PHPA on this bill
  - SB 654 – provision of information to families of children diagnosed with Down's Syndrome
    - ✓ Information needs to be given to families of Down syndrome children diagnosed both prenatally and postnatally.
    - ✓ Dr. Hoover-Fong indicated this should be a standard of care.
    - ✓ DHMH did not take a position since this is an operational impact only.
  - It was recommended that the Council members review the remaining bills for possible comments.
- **Parking for Council meeting**
  - Dr. Badawi stated that parking is no longer available in the garage for evening meetings. Department of General Services feels it is a security issue since there is not enough security in the building after 6 PM. A member of security is supposed to escort the visitor to the room from the garage. It was reported from members of the Council that they are not escorted from the front desk to the conference rooms when they come in from the outside parking lots either. The Council Chair (Anne Eder) will write a letter to the Secretary of Health to see if he can get garage parking approved.

## **VI. Next Meeting**

- Planned for April 1, 2014

**VII. Adjournment**– 8:00 PM

- Dr. Blitzer announced this is her last day on the Council. Appreciation for service expressed.  
Meeting adjourned.