

# State Advisory Council on Hereditary and Congenital Disorders

Minutes June 4, 2013

## **Members Present**

Miriam Blitzler, PhD, Chair  
Julie Hoover-Fong, MD, Co-Chair  
Anne Eder  
Delegate Shirley Nathan-Pulliam  
Coleen Giofreda  
Anika Wilkerson

## **Guests**

Carol Greene, MD

## **Staff**

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

## **Ex-Officio Present**

Fizza Majid, PhD  
Deborah Badawi, MD

## **Members Absent**

David Bromberg, MD  
Lee Woods, MD (ex-officio)  
Robert Myers, PhD (ex-officio)  
Caryl Siems  
Neal Porter, MD

**Called to Order** – 6:00 pm

## **I. Welcome and Introductions**

Members and attendees introduced themselves.

## **II. Meeting Updates**

- NBS Meeting in Atlanta
  - Report given by Dr. Majid who states over 700 attendees since International Symposium was combined with United States.
  - There was a lot of emphasis on SCID – implementation and techniques. 12 states are screening for SCID and 5 states are almost ready.
  - Lysosomal storage disorders were also discussed since Pompe has just been recommended for inclusion on recommended panel. Dr. Majid states there is no technique to test for all of LSDs at once. States that currently test for Krabbe are unable to distinguish between neonatal or adult form. It is an invasive therapy if not needed until later in life.
  - Molecular testing was also discussed in relation to CF since carriers are identified and by primary care provider is having to give this information.
  - The 2<sup>nd</sup> screen study that has been ongoing has no definite conclusions. Some 2<sup>nd</sup> screen states show more identification of CAH, but the primary disorder on 2<sup>nd</sup> screen for Maryland has been hypothyroidism.
- NYMAC Updates
  - Johnna Watson reports that the NYMAC NBS Follow-Up group has completed the baseline study for provider response in repeating NBS when requested for Unsatisfactory and Borderline specimens. Overall, response rate was around 7 days for specimen collection among all of the states. The group is now working on postcard information to send to providers emphasizing need to repeat specimens when notified.
  - Dr. Badawi reports that NYMAC had a meeting the past two days in the Baltimore Area to discuss multiple group projects.
    - They showcased two videos: one on sickle cell disease being a global disorder and another video on what families need from providers.
    - There was also discussion about consumer engagement and NYMAC's role in providing information to consumers. No definitive decision.
    - Healthcare spending – Essential health benefits in MD are equal to State Employee health plan. 60% of children are in exempt plans (large group/ERISSA) and these plans do not have to provide EHBs.

- There is also a group putting together childbirth educator education packet on NBS. They are incorporating CCHD screening as well. They are making a generic presentation and our state can make it more state-specific.

### **III. Old Business**

#### **• SCID Update**

- Update given by Dr. Majid in absence of Dr. Myers
- The lab has applied for a grant that is for implementation of SCID screening
- 3 grants are being awarded. It is unknown how many states have applied.
- The grant is for support for 2 years then the grantee should be self-sufficient.
- Award date is August 1<sup>st</sup>
- Dr Hoover-Fong reports that she had a meeting with Dr. Laura Herrera, Deputy Secretary of Health, to explain Council's purpose. It was recommended to invite her or her Chief of Staff to the next meeting and to have regular meetings with her to help keep SCID in the forefront regarding need for funding.

#### **• Sickle Cell Trait Status**

- Discussion revolved around what would be required to notify families of sickle cell trait status in MD
- Currently no notification to parents at home from the lab.
- Anika Wilkerson recommends letter to go to the home and have resource available at state for parents. However, letter could be worded to have the parent contact their baby's physician.
- Dr. Majid states we will also have to consider other trait status as well.
- It is unknown how other states currently report carrier status. Dr. Blitzer states there is currently an editorial being written which discusses how and if other states are reporting carrier status. The council would like to see this editorial.
- Johnna Watson and Dr. Badawi will ask their contacts with NYMAC if they are reporting sickle cell trait and how they are doing it, as well as what kind of follow-up or resources are available have if parents have questions.
- Will plan to gather information and discuss more in depth at September meeting.

#### **• REM diagnoses and age limits not changing:**

- Dr. Badawi reports that she spoke with Mike Berman who oversees REM and learned that the last time the list was reviewed it was a very difficult process and MA is not interested at this time in revisiting it.
- Dr. Hoover-Fong suggested development of a less political group to fix the list.
- Dr. Badawi suggests this issue may need to be revisited at a time when Dr. Herrera is in attendance.
- There was also discussion of the potential for a lot of consumer advocacy from the parent community if there are revisions on the REM list.

#### **• Health Care Reform**

- Dr Badawi provided update previously.

### **IV. New Business**

#### **• OGPSHCN Updates**

- Critical Congenital Heart Disease
  - Johnna Watson is new Program Chief for Critical Congenital Heart Disease Screening Program.
  - At a recent Advisory Council meeting it was reported that there have been some identified cases – 1 Tricuspid atresia and 1 TAPVR. There was also a Tetralogy of Fallot that passed screening but had an ECHO for a persistent murmur so was identified after screening. There has also been an interrupted aortic arch diagnosis, along with several PFO and PPHN.
  - There have also been a total of 7 parent refusals since 9/2012. Some of these same families have refused NBS as well. However, several patients had NBS later at the doctor's office but they cannot get CCHD screening there if no pulse ox is available.

- More education is needed with the hospitals to discuss lack of pulse ox in PCP office, documentation of screening and documentation of measures taken as the result of a failed screen.
- Birth Defects
  - Dr. Badawi reports that birth defects reporting system is now on-line.
  - Purpose of reporting is to identify if there are clusters of birth defects occurring which may have an environmental effect.
  - Currently, follow-up includes sending of letter to family along with resources available.
  - Dr. Badawi wants group to look at information being collected and to advise what defects should be reported. Previously, hospitals were only reporting 12 Sentinel birth defects. Now, all identified birth defects are in the new reporting system. The State currently reports information to CDC on 40 defects. Dr. Hoover-Fong states the Council along with other dysmorphologists would be willing to review and advise on what defects should be reported to the State. Dr. Badawi will send each of the lists currently used for review. This issue will be discussed at the next Council meeting.
- **Member terms expiring**
  - Dr. Blitzer's term is expiring June 30<sup>th</sup>. University of MD will be recommending replacement nomination.
  - Dr. Hoover-Fong states her term is expiring as well. She would be willing to stay for a second term. However, Dr. Hilary Vernon is also interested in being a part of the Council as well as a representative from Johns Hopkins.
  - Dr. Bromberg's term will expire so need another representative from Med Chi.
  - Anika Wilkerson's term is expiring and it would nice if another family involved with sickle cell would be interested in joining the Council
  - In general, the Council would like to have more family involvement. Anne Eder and Dr Hoover-Fong state they come into contact with families who might be interested. They would like a copy of the brochure to use in discussion with the families. A copy will be sent to them.
- **Next Meeting**
  - Will be scheduled for September 2013. A firm date was not settled at this time.

## **V. Adjournment**

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**V. Next Meetings**

- **The next meeting date is March 5, 2012.**
  
- **Topics for Future Meetings:**
  - o **Brochure**—Dr. Blitzer will send an updated brochure out for review.
  - o **Update on Blood Spot Storage and Usage** – Policy needs to be reviewed again as a routine matter every 12-18 months.
  - o **Update on NBS for lysosomal storage disorders** – Update from NYMAC lysosomal storage disorder meeting held in June.

**Adjournment** – 8:00 PM

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