

# State Advisory Council on Hereditary and Congenital Disorders

Minutes October 1, 2013

## Members Present

Miriam Blitzer, PhD, Chair  
Julie Hoover-Fong, MD, Co-Chair

## Ex-Officio Present

Fizza Majid, PhD  
Deborah Badawi, MD  
Robert Myers, PhD (ex-officio)

## Members Absent

David Bromberg, MD  
Lee Woods, MD (ex-officio)  
Caryl Siems  
Neal Porter, MD  
Anne Eder  
Delegate Shirley Nathan-Pulliam  
Coleen Giofredda  
Anika Wilkerson

## Guests

Carol Greene, MD  
Anthony Majeram, parent  
Ada Hamosh, MD  
Debbie Romanoski (Senator Dyson)  
Benjamin Smith, parent  
Kathleen Smith, parent  
Sue Blackwell, grandparent  
Hilary Vernon, MD (nominee to Council)  
Joann Kurtzberg, MD (phone)

## Staff

Johnna Watson, RN (scribe)  
Mary Dawn Celiz, NBS lab  
Donna Harris, Director, OGPSCHN  
Ilise Marrazzo, Director, Maternal Child Health

Called to Order – 6:00 pm

## I. Welcome and Introductions

Members and attendees introduced themselves.

## II. Approval of June 2013 Minutes

Minutes reviewed and approved.

## III. Special Discussion

- Newborn Screening for Lysosomal Storage Disorders
  - Several guests were in attendance to discuss adding Lysosomal Storage Disorders, such as Krabbe disease and Pompe disease, to MD's NBS screening panel.
  - Discussion was initiated by Dr. Blitzer explaining the process of how disorders are added to the MD NBS panel.
    - ✓ The Advisory Council on Hereditary and Congenital Disorders usually makes recommendations to the Secretary of Health regarding adding and modifying conditions on the panel. Decision is based on literature review, scientific input and testimony, input from all interested parties, council members, etc.
    - ✓ Legislation does not need to be passed to have conditions added.
    - ✓ Changes to the panel are implemented through regulations instead of through legislation
    - ✓ As an example, screening for cystic fibrosis was not approved initially but then it was reviewed again and was approved.
    - ✓ Screening for CCHD, as an exception, was initiated through legislation. (BUT not part of the blood spot panel)
  - Benjamin and Kathleen Smith were introduced and they presented to the Council. They explained progression of their daughter (Lily)'s condition.
    - ✓ She developed normally until 4 months of age; symptoms developed quickly and she was diagnosed in the first couple of months of age.
    - ✓ Lily had a transplant but was done after symptoms started so transplant is not as effective.
    - ✓ Mr. Smith indicates that in NY, which currently screens for Krabbe, Lily would have been transplanted prior to symptoms appearing. He provided statistics of 7 cases of LSD per year or 1:10,000 babies.
  - Anthony Majeram indicated that his child also showed symptoms at 4 months of age and passed away after 1<sup>st</sup> birthday.

- Dr. Kurtzberg (from Duke University) attended by phone and gave results of transplants that have been performed since 1995. She indicated that the earlier transplant is done, better outcome is achieved. Krabbe is most challenging to treat.
  - ✓ 16 newborns (*note from mblitzer: did not clarify what form of Krabbe disease these patients had*) have been treated secondary to family history and in utero testing.
    - 1/3 are nearly normal after 10 years
    - 1/3 are cognitively normal but some motor problems
    - 1/3 have severe motor problems but are cognitively normal.
  - ✓ Most severe forms have cognitive and motor changes in a few days of life. Dr. Kutzberg indicated that a transplant will not reverse damage. Late infantile forms will tolerate transplant at a younger age.
- NY NBS Program has screened 1.7 million babies and has identified 5 cases of early infantile – 2 are siblings. Dr. Kutzberg was asked when she planned to publish her findings because literature on screening for Krabbe disease has not been updated since 2008. Mr. Smith indicated Dr. Escolar in Pittsburgh is getting ready to publish some new data as well. The council members expressed appreciation for the presentation and support the goal of improved outcomes. Drs. Hamosh, Blitzer and Greene expressed concern over discrepancy between the information presented and the published data which shows that while the course of the disease may be slowed in some cases, neurodegeneration continues despite transplant. There was also concern expressed about the evidence for psychosocial harm to families by screening for Krabbe disease. The history of evaluation by the federal Advisory Committee on Hereditary Disorders in Newborns and Children was reviewed, including the decision to not proceed to formal evaluation of Krabbe for addition to the RUSP. It was suggested that Maryland would need to formally review the potential risks and benefits of screening for Krabbe disease in Maryland.
- It was noted by Dr. Greene that Pompe has been recommended to be included on NBS panels at the Federal level and severe combined immunodeficiency (SCID) has been recommended for several years. The Council has supported initiating screening for SCID but challenge has been funding in order to perform the testing.
- Dr. Blitzer states the Council will form a subcommittee to do information gathering on background and impacts of treatment and distribute to all council members. The subcommittee will also look at how follow-up is done in NY.

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

- CCHD Updates
  - Johnna Watson reports that screening has reached its first anniversary.
  - A relationship has been established with Children's Cardiology to determine if babies have been diagnosed after passing the screen. Working on establishing the same relationship with University of MD and Hopkins. At this time, 2 Tetralogy of Fallot and 1 TAPVR have been identified after passing the screen. However, it was discussed that Tetralogy of Fallot does not always have cyanosis (low oxygen levels) so this condition will not be picked up consistently with pulse ox screening.
  - The annual reports will be shared with the hospitals as the staff goes out to review NBS, hearing and BDRIS with the birth hospitals. The 1<sup>st</sup> sessions are planned for later this month in Western Maryland.
- Update from NBS Lab/Lab Administration
  - Dr. Myers reports that the MD NBS Lab was not awarded the grant for implementation of SCID screening.
  - He also reports that he has asked for funding for FY 15 for SCID NBS implementation. If money is appropriated in July 2014, then December 2014 would mostly likely be implementation.
  - New lab building is anticipated to be open in June 2014.
  - Council members discussed writing to the secretary and senators regarding SCID NBS.
  - It was also discussed that the support group for SCID needs to be re-engaged with this process.
- **OGPSHCN Updates**
  - Dr. Badawi reports that 3 HRSA grants have been awarded to OGPSHCN
    - ✓ Two of these grants are to improve services to children with Autism and Epilepsy through medical homes.

- ✓ The 3<sup>rd</sup> grant is for monitoring Long Term Follow-up for NBS within the medical home. Surveillance of care for children with sickle cell and hearing deficits will be completed using CRISP data.
- **NCAA & Sickle Cell Trait Pilot Project**
  - Dr. Greene reported Alexa Thomas at HRSA is the lead on this project.
  - All 3 NCAA Divisions are now requiring knowledge of sickle cell trait status for their athletes.
  - Most athletes are getting testing instead of obtaining NBS results
  - Some States will not release results secondary to screening test and not diagnostic.
  - Some coaches are calling the labs to determine information, and this is part of the medical record.

#### **V. New Business**

- **Membership Changes and Election of new Chair**
  - Dr. Hilary Vernon is being nominated by Johns Hopkins as their representative.
  - Dr. Erin Strovel, who was unable to attend tonight's meeting, is being nominated by University of MD as their representative.
  - This issue will be discussed more in-depth at the next meeting.

#### **VI. Next Meetings**

- **Topics for Future Meetings:**
  - **Brochure**—Dr. Blitzer will send an updated brochure out for review.
  - **Update on Blood Spot Storage and Usage** – Policy needs to be reviewed again as a routine matter every 12-18 months.
  - **Update on NBS for lysosomal storage disorders** – Update from NYMAC lysosomal storage disorder meeting held in June.
- Planned for 1<sup>st</sup> Tuesday of the month on a quarterly basis
  - April 1, 2014
  - June 24, 2014
  - October 7, 2014

**Adjournment** – 8:00 PM