Secretary's Advisory Committee on Heritable Disorders in Newborns and Children

Summary of 30th Meeting
April 19, 2013
Webinar

The Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) was convened for its 30th meeting at 10:00 a.m. EST and adjourned at 4:01 p.m. EST on Friday, April 19, 2013, as a webinar. In accordance with the provisions of Public Law 92-463, the meeting was open for public comment.

COMMITTEE MEMBERS

Don Bailey, Ph.D., M.Ed.
Distinguished Fellow
Early Childhood Development
RTI International
3040 East Cornwallis Road
P.O. Box 12194
Research Triangle Park, NC 27709-2194
Phone: (919) 541-6488
Email: dbailey@rti.org

Joseph A. Bocchini, Jr., M.D.
(Committee Chairperson)
Professor and Chairman
Department of Pediatrics
Louisiana State University
Health Sciences Center in Shreveport
1501 Kings Highway
Shreveport, LA 71103
Phone: (318) 675-6073
Email: jbocch@lsuhsc.edu

Jeffrey Botkin, M.D., M.P.H.
Professor of Pediatrics and Medical Ethics
Associate Vice President for Research
University of Utah
Research Administration Building
75 South 2000 East #108
Salt Lake City, UT 84112-8930
Phone: (801) 581-7170 or 7171
Email: jeffrey.botkin@hsc.utah.edu

Charles Homer, M.D., M.P.H.
Chief Executive Officer and President
National Initiative for Children's
Healthcare Quality
30 Winter Street, 6th Floor
Boston, MA 02108-4720
Phone: (617) 391-2702
Email: chomer@nichq.org

Fred Lorey, Ph.D.
Acting Chief
Genetic Disease Screening Program
California Department of Public Health
850 Marina Bay Parkway
Richmond, CA 94804
Phone: (510) 412-1490
Email: fred.lorey@cdph.ca.gov

Dietrich Matern, M.D.
Department of Laboratory Medicine
and Pathology
Mayo Clinic
200 First Street S.W.
Rochester, MN 55905
Phone: (507) 538-1581
Email: dietrich.matern@mayo.edu

Stephen McDonough, M.D.
Medicenter One Health Systems, Inc.
Department of Pediatrics
222 7th Street, N
P.O. Box 5505
Bismarck, ND 58502-5505
Phone: (701) 323-5355
Email: smcdonough@mohs.org

Alexis Thompson, M.D.
Division of Hematology/Oncology
Children's Memorial Hospital
2300 Children's Plaza Box #30
Chicago, IL 60614
Phone: (312) 227-4834
Email: athompson@luriechildrens.org

Catherine A. L. Wicklund, M.S., C.G.C.
Northwestern University
Feinberg School of Medicine
Center for Genetic Medicine
676 N. St. Clair, Suite 1280
Chicago, IL 60611
Phone: (312) 926-7468
Email: c-wicklund@northwestern.edu

Andrea M. Williams, B.A.
The Children's Sickle Cell Foundation, Inc.
617 Gearing Avenue
Pittsburgh, PA 15210
Phone: (412) 853-9883
Email: awilliams@cscfkids.org

EX-OFFICIO MEMBERS

Agency for Healthcare Research and Quality
Denise Dougherty, Ph.D.
Senior Advisor
Child Health and Quality Improvement
540 Gaither Road
Rockville, MD 20850
Phone: (301) 427-1868
Email: ddougher@ahrq.gov

Centers for Disease Control and Prevention
Colleen A. Boyle, Ph.D., M.S.
Director
National Center on Birth Defects and
Developmental Disabilities
1825 Century Center Boulevard
Mailstop E86
Atlanta, GA. 30345
Phone: (404) 498-3907
Email: cab3@cdc.gov

Food and Drug Administration
Kellie B. Kelm, Ph.D.
Scientific Reviewer/Biologist
Division of Chemistry and
Toxicology Devices
Office of In Vitro Diagnostic Devices
Evaluation and Safety
10903 New Hampshire Avenue
WO66, Room 5625
Silver Spring, MD 20993-0002
Phone: (301) 796-6145
Email: kellie.kelm@fda.hhs.gov

Health Resources and Services Administration
Michael Lu, M.D., M.P.H.
Associate Administrator
Maternal and Child Health Bureau
5600 Fishers Lane, Room 18-05
Rockville, MD 20857
Phone: (301) 443-2170
Email: mlu@hrsa.gov

National Institutes of Health
Alan E. Guttmacher, M.D.
Director
Eunice Kennedy Shriver National Institute of
Child Health and Human Development
31 Center Drive
Building 31, Room 2A03
Bethesda, MD 20892-2425
Phone: (301) 496-3454
Email: guttmach@mail.nih.gov

DESIGNATED FEDERAL OFFICIAL
Debi Sarkar. M.P.H.
Health Resources and Services Administration
Genetic Services Branch
Maternal and Child Health Bureau
5600 Fishers Lane, Room 18A-19
Rockville, MD 20857
Phone: (301) 443-1080
Email: dsarkar@hrsa.gov

LIAISONS AND ORGANIZATIONAL REPRESENTATIVES

American Academy of Family Physicians
Frederick M. Chen, M.D., M.P.H., F.A.A.F.P.
Family Practitioner
Department of Family Medicine
University of Washington
4311 11th Avenue, NE, Suite 210
Seattle, WA 98195-4982
Phone: (206) 543-7813
Email: fchen@u.washington.edu

American Academy of Pediatrics
Beth Tarini, M.D., M.S., F.A.A.P.
University of Michigan Health System
300 N. Ingalls Street 6C11
Ann Arbor, MI 48109
Phone: (734) 223-4416
Email: btarini@umich.edu

American College of Medical Genetics
Michael S. Watson, Ph.D., F.A.C.M.G.
Executive Director
9650 Rockville Pike
Bethesda, MD 20814-3998
Phone: (301) 634-7127
Email: mwatson@acmg.net

American College of Obstetricians and Gynecologists
Nancy C. Rose, M.D.
Chair, ACOG Committee on Genetics
Director, Reproductive Genetics
Intermountain Healthcare
5121 S. Cottonwood Street
Salt Lake City, UT 84157
Phone: (801) 507-7431
Email: nancy.rose@hsc.utah.edu

Association of Maternal and Child Health Programs
Lisa Bujno, M.S.N., A.P.R.N.
Chief, Bureau of Public Health Systems, Policy, and Performance
New Hampshire Division of Public Health Services
29 Hazen Drive
Concord, NH 03301
Phone: (603) 271-4110
Email: lbujno@dhhs.state.nh.us

Association of Public Health Laboratories
Susan M. Tanksley, Ph.D.
Manager, Laboratory Operations Unit
Texas Department of State Health Services
P.O. Box 149347
Austin, TX 78714
Phone: (512) 776-3106
Email: susan.tanksley@dshs.state.tx.us

Association of State and Territorial Health Officials
Christopher Kus, M.D., M.P.H.
Associate Medical Director
Division of Family Health
New York State Department of Health
Corning Tower, Room 2162
Albany, NY 12237
Phone: (518) 473-9883
Email: cak03@health.state.ny.us

Genetic Alliance
Natasha F. Bonhomme
Vice President of Strategic Development
Genetic Alliance
4301 Connecticut Avenue, NW, Suite 404
Washington, D.C. 20008-2304
Phone: (202) 966-5557 Ext. 211
Email: nbonhomme@geneticalliance.org

March of Dimes
Edward R.B. McCabe, M.D., Ph.D.
Senior Vice President and Medical Director
March of Dimes
1275 Mamaroneck Avenue
White Plains, NY 10605
Phone: (914) 997-4649
Email: emccabe@marchofdimes.com

National Society of Genetic Counselors
Cate Walsh Vockley, M.S., C.G.C.
Senior Genetic Counselor
Division of Medical Genetics
Children's Hospital of Pittsburgh
4401 Penn Avenue
Pittsburgh, PA 15224
Phone: (412) 692-7349
Email: catherine.walshvockley@chp.edu

Society for Inherited Metabolic Disorders
Carol Greene, M.D.
University of Maryland Medical System
Pediatric Genetics
737 West Lombard, Room 199
Baltimore, MD 21201-1596
Phone: (410) 328-3335
Email: cgreene@peds.umaryland.edu

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I. Committee Business: April 19, 2013

A. Welcome and Roll Call

Joseph A. Bocchini, Jr. M.D.
Committee Chair
Professor and Chairman
Department of Pediatrics
Louisiana State University
Shreveport, LA

Dr. Bocchini welcomed the webinar participants and offered instructions on how to attend an Advisory Committee meeting in webinar format. He then proceeded to take roll for the thirtieth meeting of the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC). Voting members present were: Dr. Don Bailey, Dr. Colleen Boyle, Dr. Denise Dougherty, Dr. Alan Guttmacher, Dr. Charles Homer, Dr. Kellie Kelm, Dr. Fred Lorey, Dr. Michael Lu, Dr. Stephen McDonough, Dr. Dietrich Matern, Dr. Alexis Thompson, Ms. Catherine Wicklund, and Ms. Andrea Williams. Ms. Debi Sarkar served as the Designated Federal Official (DFO).

Nonvoting organizational representatives participating in the webinar were:

- American Academy of Pediatrics: Dr. Beth Tarini
- Association of Public Health Laboratories (APHL): Dr. Susan Tanksley
- Association of State and Territorial Health Officials: Dr. Christopher Kus
- Genetic Alliance: Ms. Natasha Bonhomme
- March of Dimes: Dr. Edward McCabe
- Society for Inherited Metabolic Disorders: Dr. Carole Greene

B. Approval of January 2013 Minutes

Committee members offered no comments on the minutes of the SACHDNC's January 2013 meeting. Dr. Bailey made a motion to approve the January minutes, which was seconded by Dr. Boyle. All of the committee members present voted to approve the minutes.

C. Update on Committee and Other Committee Business

Dr. Lu thanked the Committee members for their ongoing leadership and service. He reminded the call participants that the SACHDNC's charter expires on April 24 and reported that Secretary of Health and Human Services (HHS) Kathleen Sebelius decided to create a discretionary committee to carry out the functions currently performed by the SACHDNC. There are several logistical steps, including the posting of a notice in the *Federal Register*, that must be completed before the new Committee can be formally created. He anticipated that these administrative steps would be completed in time for the next meeting, which is scheduled to take place on May 16-17. Dr. Lu promised to share more information on the new Committee as soon as it is available.

Dr. Lu announced that Ms. Sarkar has taken over as the DFO for the SACHDNC. Ms. Sarkar has worked with the Committee for the past year and has extensive experience with newborn screening (NBS) issues and with the Health Resources and Services Administration (HRSA). Ms.

Sarkar greeted the webinar participants and stated that her team is working diligently to set up the new Committee.

Dr. Bocchini added that the SACHDNC members were very pleased to know that they would be able to continue their work under the new discretionary committee. He also thanked the various organizations that support the work of the SACHDNC and welcomed Ms. Sarkar on behalf of the SACHDNC members.

D. Approval of the 2012 Annual Report to Congress

Dr. Bocchini thanked the SACHDNC staff for writing a very strong annual report that illustrates the breadth of the Committee's work in 2012. The report describes the SACHDNC's mission, provides an executive summary, summarizes the Committee's activities, and includes a forecast of future activities organized by the priorities of the Committee and the charges given to the three SACHDNC subcommittees. Dr. Bocchini reviewed the Committee's accomplishments included in the report and noted the extensive work done by the Subcommittees.

None of the Committee members offered comments or recommendations concerning the report. Dr. Bailey made a motion to accept the report as written. Dr. McDonough seconded the report. The motion passed with the unanimous consent of the voting members present.

Committee Discussion

- Dr. McDonough asked whether the May meeting of the new discretionary committee would be a virtual meeting (webinar) or an in-person meeting. Dr. Bocchini anticipated that it would be a webinar. Ms. Sarkar promised to share the details of the May meeting once the discretionary committee is established.

II. Update on RUSP Conditions

A. Lessons Learned from Early Hearing Detection and Intervention (EHDI) that May Be Applicable to Critical Congenital Heart Disease (CCHD) Screening – Update

Christopher Kus, M.D., M.P.H.
Associate Medical Director
Division of Family Health
New York State Department of Health
Albany, NY

Dr. Kus reported that the Follow-Up and Treatment Subcommittee has been looking into ways to apply the lessons learned from early hearing detection and intervention (EHDI) efforts to Critical Congenital Heart Disease (CCHD) screening efforts. He highlighted the major points on which the Subcommittee will report. The Subcommittee anticipates presenting a final paper to the SACHDNC members prior to the September meeting and asking for the Committee's approval of the report during the meeting.

Major lessons learned were:

- State EHDI and newborn bloodspot programs are often not well integrated with each other. As the number of point-of-care screenings increase, it will be more and more important that public health NBS programs are integrated.

- State health departments should play a leadership role in implementing electronic data systems that utilize standards-based messaging to reduce errors and enhance timely data reporting. EHDI serves as a model of electronic information exchange between clinical care and public health programs. The goal is to leverage the extensive work currently underway to facilitate the electronic transfer of NBS results between the laboratory and the primary care physician.
- As new tests are added, appropriate state and federal financial support will be needed to develop the CCHD screening system, including funding for implementation of follow-up systems.
- Screening systems should require child-level data, including follow-up information, to support quality improvement efforts.
- Appropriate state and federal financial support will be required to integrate CCHD screening into existing data systems or to enhance interoperability among NBS systems when integration is not possible.

Committee Discussion

- Dr. Lorey pointed out that it is not possible to integrate hospital-based and laboratory testing in some states (e.g., these functions are in different departments). He was not convinced that the two types of testing should be integrated, although he was in agreement with Dr. Kus' comments on funding issues. Dr. Kus acknowledged that there are differences of opinion concerning this issue. He believed that NBS reporting should be consistent across all screenings and include all screenings. Dr. Kus anticipated that there would be greater integration of the point-of-care screenings and promised that the Subcommittee would take comments such as these into account as it prepares the final report.
- Dr. Homer emphasized the importance of the Committee's recommendations concerning the screening of all children and the provision of appropriate follow-up. The Committee should stress the importance of the overarching public interest of ensuring appropriate screening. Dr. Kus agreed that the Committee should support the dissemination of these types of messages.
- Dr. Bocchini asked how many states have the type of separation within public health functions described by Dr. Lorey. Dr. Kus indicated that the Subcommittee conducted a survey that included this information and indicated that it could be added to the report. He noted that EHDI tends to be linked to early intervention programs instead of bloodspot screening programs, which results in the separation. Each state deals with the relationship between the screenings differently. The Subcommittee could include this information in the report.
- Dr. Bocchini asked whether the Subcommittee has identified any potential cost savings associated with overlaps in the administrative structure of the screening programs. Dr. Kus indicated that one area that could potentially provide savings is follow-up mechanisms. He expressed interest in determining whether EHDI could take advantage of existing bloodspot screening follow-up mechanisms to increase efficiency. He was uncertain whether savings could be found in long-term follow-up since it has not been funded in the past; however, a coordinated model should be less expensive.
- Dr. Boyle identified administrative savings as an important issue and noted that two of the lessons learned address financial support for aspects of CCHD screening. She suggested that the Subcommittee compare two different state models and illustrate the effects of integration and long-term follow-up and emphasize the importance of integrating new screenings into existing programs as they are added. Dr. Kus anticipated that the Subcommittee would highlight state screening collaboration models that could be applied to CCHD screening.
- Dr. Bocchini asked whether there is enough cost data from the individual states to allow the Subcommittee to determine cost per patient. Dr. Kus did not know if there was enough data available to undertake such an estimate. The CCHD grant included funds to

collect cost data. He did not think that there was dedicated funding for collecting EHDI costs.

- Dr. Greene expressed her concerns about how deeply into the process the Subcommittee would be willing to go with regard to long-term data and the ramifications of its recommendations. Costs for data collection will vary by state. States that already have plans for integrating NBS data regardless of source will look at costs very differently than those that will need to build a system to bring the data together. She pointed out that there is a significant difference between following up on babies with abnormal pulse oximeter readings compared to following up on all babies with conditions that the SACHDNC has been discussing (except thyroid). There are also questions associated with follow-up for babies with heart defects that were not picked up by a screening because they were not critical cyanotic congenital heart defects. Follow-up could become very complicated if all heart defects are tracked as it would mean tracking 1 out of every 100 babies. Dr. Greene emphasized the importance of giving careful consideration to the way that long-term data in the heart population could be tied to screening questions.
- Ms. Kimberly Piper commented via the chat function of the webinar that another lesson learned was that CCHD was recommended to states for addition to their screening panels without examination of the capacity of systems to implement the screening, which is very different from the established processes states already use. Dr. Kus responded that a public health infrastructure statement has been added to new reviews of conditions. The SACHDNC learned from the CCHD experience and is working to incorporate these lessons into its ongoing work.
- Dr. Tanksley reported that the APHL's Newborn Screening Technical assistance and Evaluation Program (NewSTEPs) is collecting information concerning the overlap between EHDI and bloodspot screening programs and anticipated that APHL would be able to make this information available to the Subcommittee for use in its report.

B. Assessing the Impact of the NCAA Sickle Cell Trait Screening on State Newborn Screening Programs

Beth Tarini, M.D., M.S., F.A.A.P.
University of Michigan Health System
Ann Arbor, MI

Annie Azrak
Research Assistant
Child Health Evaluation and Research (CHEAR) Unit
Division of General Pediatrics
University of Michigan
Ann Arbor, MI

Dr. Tarini stated that her presentation would address a project that is one part of a two-part contract between the Genetic Alliance and HRSA to assess the policy impact of non-disorder related recommendations, the factors that prevent states from sharing sickle cell trait (SCT) screening results, and the effect of the SACHDNC's recommendations on states. She anticipated presenting the results of the second part of the project in the near future. Dr. Tarini reviewed the history of the requirement, the content of the SACHDNC's recommendation, the potential impacts of the requirement on NBS programs (i.e., demand on resources, programmatic changes, and variation of impact), the methodology of the study, the history of NBS SCT screening, the effects/burdens of the mandate on NBS screening programs, and the results to date.

The National Collegiate Athletic Association (NCAA) SCT screening mandate, which went into effect in 2010, requires all Division I and Division II athletes to have a sickle cell solubility test as part of their pre-participation medical evaluations or to provide a prior test result. Athletes also

have the option of signing a waiver and foregoing the test. In early 2013, the NCAA implemented a sickle cell testing requirement for Division III athletes.

The project used a phone survey and a snowball sampling method to interview laboratory directors and personnel, follow-up directors and personnel, hematologists, genetic counselors, and sickle cell community-based organizations. The interviews addressed the history and procedures for screening used in each state and the direct effects of the NCAA mandate. To date, the researchers have contacted 86 percent of the states and completed work on 24 states.

Dr. Tarini summarized the conclusions of the research based on the work done to date:

- Not all states are able to provide the result of SCT screening to student athletes, even though SCT is part of the NBS screening program.
- Practices concerning the sharing of SCT screening results vary from state to state.
- There is great variability in the impact of the mandated sharing of SCT screening results among the states.

Committee Discussion

- Ms. Wicklund asked whether the researchers obtained data concerning the number of requests for SCT screening results prior to the mandate or whether they only obtained data on the number of requests since the mandate went into force. Ms. Annie Azrak replied that the researchers only received information on the number of requests received since the passage of the mandate. In follow-up questions, the researchers asked whether the number of annual requests reflected an increase and, if so, which year the increase began. With the exception of one state, all responding states reported that SCT result requests had increased beginning in 2010, but some states did not see an influx of requests until 2012.
- Ms. Williams asked whether the researchers collected information on the type of educational information states sent out along with the requested results. Ms. Azrak indicated that some states send out the same brochure, containing information about SCT and where to go for more information, that they would send to any family that received an SCT diagnosis whether or not they had a record to provide. This generally correlated with the state's NBS screening reporting procedure. Some states referred requesters to other resources for more information. A final group of states did not provide any information. Dr. Tarini added that some states were unhappy because proposed partnerships to develop educational programs with the NCAA and athletic programs did not come to fruition.
- Dr. Thompson asked for more information concerning the purpose of the project and the direction in which it is heading. She was also concerned that the Committee was focusing on the NCAA when its obligation is to the general public, many of whom would be interested in learning their carrier status. Dr. Tarini stated that the project is not a comment on the medical necessity or medical legitimacy of the mandate, nor is it an endorsement of the mandate or the NCAA's decision to promulgate it. The project is focused on the larger issues of the public health impact that accompany the SACHDNC recommendations concerning disorders and their impact on state NBS programs. Dr. Tarini said that the work raises larger questions concerning the duty and feasibility of providing past and future test results and education to all carriers and providing test results for non-carriers who were previously screened.
- Ms. Williams expressed her concern about the unintended harms to the individual that might result from the patchwork of educational efforts undertaken by the states. The student athletes might or might not have received information on their genetic information, on its meaning, and on how it should be protected. Dr. Tarini disagreed that the problems are as great as Ms. Williams believed. When states provide results, they follow their protocols for providing information on SCT in general. With regard to the potential harms to individual athletes and the education about SCT they receive, Dr. Tarini believed, but did not have confirming data, that the vast majority of athletes

receive their results from their institutions. The issue of education should be addressed, but should be addressed by the universities and athletic departments. The research shows that some states are feeling a burden on resources and time based on the need to provide results, but the vast majority of athletes are obtaining this testing through other means than the states.

- Ms. Wicklund stated that this situation illustrates the impact that a mandate by an outside body can have on the states. It also raises questions about the responsibility of the state to address this issue, especially when it takes resources from other necessary functions. Dr. Tarini agreed that the issues of the state's responsibility and of optimal counseling are important to discuss. Ms. Azrak added that the snowball sampling method asks for the names of hematologists, sickle-cell community-based organizations, and genetic counselors. Some of the questions that are asked during these subsequent interviews address the types of genetic counseling offered within each state, the typical procedure for directing families with a positive SCT result through genetic counseling, and the involvement of sickle-cell organizations in the educational efforts.
- Dr. Kus noted that the SACHDNC came out with a recommendation based on good science. He stated that the NCAA's policy, which came out before the Committee's, is not based on good science. He asked whether the Committee now has a role to play in questioning the need to continue the NCAA mandate. Dr. Bocchini believed that the question was a good one, but indicated that there was a need for greater discussion of it within the Committee.
- Dr. McCabe asked whether the specificity and sensitivity of NBS for SCT is known overall and by state. He asked whether the Centers for Disease Control and Prevention (CDC) has this type of data in its quality assurance program. Dr. Tarini replied that she does not have the data he asked for. She indicated that the states do not give out the results unless they are certain that they can stand by them.
- Dr. Tanksley stressed that NBS is not a diagnostic test. Without diagnostic confirmation, there is no way to be certain that an individual has a particular condition. Transfusions can interfere with the results of certain screenings. Ms. Azrak added that some states mentioned providing a cover letter along with the results that emphasizes that the results are only from a screening and are not a diagnosis.
- Dr. Sara Copeland explained that the purpose of this work was to determine the impact of SACHDNC policy statements such as the one on SCT screening. It seems as though there was some amelioration concerning the NCAA recommendation with regard to education but not about who should be screened (i.e., everyone). The HRSA funded project was not supposed to be an endorsement or a condemnation of the NCAA mandate; its purpose was to determine the impact of the Committee's recommendation and whether states could use it to frame their response to the mandate. Dr. Tarini observed that states might have a conflict between the federal Committee's recommendations and overriding individual state policies concerning the release of results. She did not believe that states could refuse records requests if they have the results and believe them to be valid.
- Dr. Copeland stressed that this issue resides at the state level. All the SACHDNC can do is support the states as best it can by providing tools, resources, and guidelines. Technically, outside groups should have little or no impact on NBS programs, but, in this case, the NCAA has. Dr. Tarini noted that the focus of the research was how the states are dealing with the requests generated as a result of the NCAA mandate, even though the mandate does not align with the SACHDNC's recommendations.
- Dr. Thompson asked whether problem solving efforts by states associated with the NCAA mandate benefit the public overall. Dr. Tarini said that her discussions with the states indicate that they are committed to handling this issue in the best possible way. States have tried to partner with NCAA athletic programs. Dr. Thompson was troubled that the response seemed focused on the athletic departments, not the public and the public good. She was particularly concerned that athletes could sign a waiver of liability against an institution, which does not indicate that the health of the individual athlete is the primary concern of the institution. Solutions should focus on a wider range of people,

such as pregnant women with babies at risk for sickle cell, rather than on a narrow group like an athletic department. Dr. Tarini reported that she has, as part of the project, reached out to other stakeholders (e.g., athletic departments, the NCAA, and the National Institutes of Health) to request that they consider the policy terms of the original litigation and the medical legitimacy of the mandate, and work together to educate the athletes. It is not optimal to provide the results to the institutions without a plan for educating their students.

- Dr. Thompson believed that carrier education conducted in direct response to the mandate could potentially help the broader community. Dr. Tarini stated that the mandate offers an opportunity to create a model for education that could be broadly disseminated and used for SCT counseling.
- Dr. Greene expressed concerns about the effects of the shift toward DNA screening, long-term follow-up for identified carriers, and the capacity of states to take on additional responsibilities. Ms. Williams pointed out the need for more information concerning follow-up for the thousands of individuals who have already received their results and for those who will receive them.
- Dr. Kus wondered how many athletes in the greater population collapse during, or as a result of, practice. Dr. Tarini noted that SCT is one of several reasons an athlete could collapse or die during intense conditioning. She supported mandating prevention measures, not just screening.
- Noting that the central question seems to be the role of the Committee with regard to this issue, Dr. Bocchini thought that it was within the group's purview to reiterate its position on the routine screening of athletes. He thought the Committee should also raise the issue of data being provided without proper education and follow-up as well as the potential for misuse of screening data. He asked whether the members thought that the Committee should develop a document or study to highlight the additional problems that are developing. Dr. Tarini stressed that the results she reported were preliminary results.
- Dr. Tarini estimated that she would be able to have a complete data set by June 1. Dr. Bocchini noted that this timeline would enable the Committee to review the data prior to the September meeting and make decisions about how to proceed at that time (e.g., develop additional data to highlight the problems the mandate created). The resulting data could be used to educate the state public health departments on how to address these issues. Dr. Tarini supported this approach, especially since it would identify the core elements that need to be addressed in the process for all stakeholders. This would also serve as a good follow-up to the Committee's recommendation. Dr. Bocchini pointed out that the only problem that the survey does not address is the potential negative effects for the individual athletes. Dr. Tarini indicated that it is difficult to gain access to the athletes.

C. Newborn Screening for CCHD: An Update on CDC Activities

Cynthia F. Hinton, Ph.D., M.S., M.P.H.
Health Scientist
Pediatrics and Genetics Team
Division of Birth Defects and Developmental Disabilities
Centers for Disease Control and Prevention
Atlanta, GA

When Secretary Sebelius endorsed the SACHDNC's recommendation for CCHD screening, she charged the CDC with three tasks: 1) evaluate state surveillance and tracking to monitor the effectiveness of CCHD NBS; 2) conduct a cost-effectiveness analysis of NBS for the early identification of CCHD; and 3) leverage an electronic health record framework for congenital

heart defects (CHD), including CCHD. Dr. Hinton presented an update on the CDC's efforts in each of these areas.

Dr. Hinton described the CDC's efforts to support surveillance, public health practice, and applied research. These efforts include surveying states about their perceived role with regard to CCHD screening and any anticipated problems regarding the screening, studying the role of birth defects surveillance programs in evaluation within state CCHD screening programs, providing direct aid (staffing) to states through the CDC's Epi-Aid program, supporting birth defects surveillance programs, and providing technical assistance to the Nation Birth Defects Prevention Network (NBDPN). Currently, CDC funds 14 state programs and, in conjunction with the NBDPN, provides technical assistance to states concerning the development and enhancement of surveillance systems and collaborates on epidemiologic analyses. Each year, the NBDPN publishes an annual report on 41 birth defects. In 2012, the report focused on CCHDs, including their prevalence by state and surveillance system. CDC also supported a one-day Association of Maternal and Child Health Programs (AMCHP) meeting, focused on interactions between birth defect surveillance organizations and state NBS programs that emphasized improving communications and breaking down barriers to cooperation. The CDC has access to the Metropolitan Atlanta Congenital Defects Program, which is an active, state and population-based surveillance program. Data from the program can be linked to other types of data to identify survival trends over a long period of time. The CDC supports the applied research conducted by the National Birth Defects Prevention Study, including an effort to determine the proportion of cases of CCHD that might benefit from the new recommendations concerning NBS for CCHD. Another project supported by the CDC is the Florida March of Dimes research, comparing mortality and hospital resource utilization among infants with timely and late CCHD detection.

With regard to health economics, CDC supported three studies that are currently in the peer review process. A study in New Jersey looked at data reporting costs and the costs associated with conducting screening (i.e., time-motion studies of pulse oximetry screening). A Florida study examined service utilization and costs for late diagnosis. The final study looked at overall cost-effectiveness for routine CCHD NBS.

Dr. Hinton also noted that the Agency for Healthcare Research and Quality's Healthcare Cost and Utilization project is examining discharge-level hospital administrative billing data to determine the health care resource utilization of pediatric and/or adult congenital heart defect hospital discharges at different ages, how these discharges differ in their health care utilization from discharges with non-critical heart defects, and which factors affect the healthcare utilization of the discharges with CHDs, including CCHDs.

A cross-agency effort on leveraging electronic health records with the National Library of Medicine (NLM) and NHLBI is focused on mapping CCHD conditions to various coding systems. The goal of the project is to facilitate meaningful data exchange between the various stakeholders. An abstract on this work will be presented at the NBS meeting in Atlanta in May.

Committee Discussion

- Ms. Bonhomme asked whether educational efforts were discussed during the AMCHP meeting. Dr. Hinton replied that the subject was not discussed during the meeting; the meeting focused on the activities of birth defects surveillance programs and NBS programs, and the effects of Title V on helping them work together.
- Ms. Bonhomme asked whether there had been discussions concerning educating the parents of identified children or the general public about CCHD screening. Dr. Hinton indicated that the CDC has an educational webpage concerning CCHD. She also believed that the staff within her Center works closely with the birth defects surveillance programs to help connect affected families with services and education. The analyses done by her group do not focus on the educational aspect. She indicated that the Heart Defect Collaborative might be conducting educational activities.

- Dr. Kus asked about the level of involvement of the malformation registry programs with the implementation grantees. Dr. Hinton replied that it depends on where the birth defects program is located within the state structure and the way in which the communication systems are set up. Having agreements concerning the sharing and merging of data is more important that where the two entities are housed. Birth defects programs do not operate with the same rapidity as the bloodspot screening programs, but they are very good at obtaining patient-level data in a timely manner. Dr. Kus added that New York's congenital malformation registry was not a big player in the implementation of the hearing screening, but it has been helpful with regard to long-term follow-up.
- Dr. Boyle suggested that the Committee should follow the early implementers, identify the challenges they faced, and determine how it can help as implementation continues. Dr. Hinton stated that CDC began a pilot project last year in several states to conduct surveillance for CHDs in adolescents and adults and determine how well they do over their life courses.
- Dr. McCabe suggested that the CDC look into working with the clinics that serve adults with CHDs. He believed that there are several consortia of these clinics nationwide. Dr. Hinton believed that they are being included in the project.

General Discussion

- Mr. Harry Hannon asked via the chat function of the webinar whether loss to follow-up for CCHD is better than loss to follow-up for hearing loss screening. Dr. Hinton indicated that it is probably too early to answer this question. She pointed out that CCHD differs from hearing loss in that the infants are critically ill and require immediate specialty supervision, which minimizes the immediate loss to follow-up. She indicated that she did not have information on long-term follow-up after the immediate crisis.
- Dr. Alan Zuckerman commented, via the chat function, that the Society of Thoracic Surgeons might be able to provide data on follow-up for CCHD across state lines and on long-term outcomes.

III. Public Consideration

A. Pompe Condition Nomination – Update

Joseph A. Bocchini, Jr. M.D.
Committee Chair
Professor and Chairman
Department of Pediatrics
Louisiana State University
Shreveport, LA

Dr. Bocchini indicated that the Condition Review Workgroup (CRW) would present its findings concerning Pompe disease during the May meeting. The CRW was in the process of finalizing the evidence review, completing the decision analysis process, and conducting the assessment of the public health impact.

B. Public Comment

Dean Suhr, Advocate, MLD Foundation: Mr. Suhr reiterated his interest in discussing the possibility of changing the criteria for SACHDNC approval of NBS so that it no longer requires a viable therapy as a criterion for recommendation. The implications of changing the criteria are significant in terms of philosophy, focus, work, and the cost of social and medical services. Many

organizations are interested in the consequences of knowing that a child has a potentially fatal or very serious disease in advance of the symptoms. He hoped to be able to convene a caucus on this issue in the next 6 to 12 months. Mr. Suhr requested that the Education and Training Subcommittee consider creating or making available materials that help advocacy groups to work with families and organizations that are advocating for the adoption of screenings and volunteered to help with the development of these materials.

Amber Salzman, President, Stop ALD Foundation: Dr. Salzman reminded the participants that the SACHDNC reviewed the ALD NBS nomination during its September 2012 meeting, determined that it is a medically-important disorder that merits serious consideration, and requested more prospective data from the Mayo Clinic pilot study. She provided a brief update on the Mayo study, which has analyzed 50,000 samples from California newborns and plans to complete a similar number by the end of September. The ALD screen used in the study has a very low false positive rate, correctly identified all of the control samples in a blinded study, and has existing mechanisms to conduct molecular screenings on samples that test positive on the biochemical screen. Dr. Salzman requested that the Committee provide guidance on the best ways to move the review of ALD forward in an expeditious manner. She also reported that New York voted to implement ALD screening and required that the test be validated in the laboratory in time to begin screening in January 2014.

IV. The Affordable Care Act and the Impact on Individuals with Heritable Disorders

Meg Comeau, M.H.A.
Project Director
The Catalyst Center
Health and Disability Working Group
Boston University School of Public Health
Boston, MA

Ms. Comeau began by reviewing the intersection between the public health system, which focuses on improving population health, and insurance coverage, which focuses on reducing individual financial risk, and the way in which insurance has become more important to keeping the NBS system viable as state and federal funding for public health has become tighter. Getting and keeping individuals on publically or privately funded affordable coverage increases access to broader, long-term access to care and leads to opportunities for improved individual, and ultimately population, health.

The Affordable Care Act (ACA) is a combination of two laws passed in March 2010. The ACA includes several provisions of interest to those with heritable conditions. It prohibits the denial of coverage to individuals with pre-existing conditions, expands dependent coverage to children up to age 26, and prohibits rescission of coverage. Beginning in 2014, it requires guaranteed issue and guaranteed renewal and prohibits discrimination based on health status, including genetic information. The ACA eliminates lifetime benefit caps and requires annual benefit caps to exceed $2 million through January 2014, when annual caps will be eliminated.

Ms. Comeau described the state marketplaces that will open in January 2014, including eligibility rules, assistance available for enrolling, and available tax credits and subsidies. Plans in the marketplace must offer a core group of Essential Health Benefits (EHBs). Large group plans and grandfathered plans, the type of plans through which most children with special health care needs and heritable conditions are insured, are exempt from the EHB requirement. The EHB definition cannot be used to discriminate against individuals because of age, disability, or expected length of life. The 10 EHBs include wellness and chronic disease management services. They are

determined on a state-by-state basis using a benchmarking process which allows states to identify the most appropriate, cost-effective, and helpful benchmark options. States can mandate benefits that go beyond the scope of EHBs.

The expansion of the Medicaid program also provides a new pathway to coverage. The expansion is optional for states. It expands the eligible population to include non-disabled, non-pregnant adults and increases the income level for eligibility. The new income eligibility does not apply to children; however, eligible income levels for this population will increase in 2014 to match that of the newly eligible population.

The ACA also includes provisions addressing cost and quality. The law increases the Medicaid primary care reimbursement rate to the same level as the Medicare rate in hopes of increasing the pool of providers who will accept Medicaid. Accountable care organizations (ACOs) will function as medical home "neighborhoods" linking a network of hospitals and specialists responsible for cost and quality across the care continuum. Ms. Comeau stressed the importance of including individuals with expertise in clinical pediatrics and genetics in the development and design of ACOs to ensure that the patient populations, the focus of the Committee, are built into the system from the beginning. Section 2703 provides a means of funding, through the Medicaid State Plan Amendment (SPA) process, select operational components of medical homes (e.g., care coordination for pediatrics) for a specifically defined group of patients that generally do not have funding available in traditional private insurance or Medicaid. Services and supports included under Section 2703 include care management and coordination, individual and family support, and referral to community and social support services. The provision also includes enhanced federal reimbursement for services covered under the program.

Ms. Comeau also noted that the ACA requires all new employer-sponsored or individual plans/policies to include several preventative services without co-pays, co-insurance, or deductibles being charged. Some of the services in this group that are relevant to genetics include screening and counseling for women at high risk of breast cancer, congenital hypothyroid screening for newborns, hemoglobinopathies or sickle cell screening for newborns, phenylketonuria NBS, autism and developmental screening, and newborn metabolic and hemoglobin screening. Fully insured and self-funding plans are required to provide coverage for screenings included on the Recommended Uniform Screening Panel (RUSP), without cost-sharing, regardless of whether the state has adopted the complete panel or only specific conditions on the RUSP.

Ms. Comeau concluded her remarks by stating that the ACA offers historic opportunities for individual insurance coverage and moving the field of public health forward; however, the Act does not solve all problems. Issues that still need to be addressed include the fact that all of the law's provisions do not apply to every kind of private and public health insurance coverage and that the EHBs are being built upon existing coverage models, which could perpetuate coverage gaps for those with heritable disorders. Additionally, the long-term sustainability of funding at both the state and federal levels remains a concern as does the continued need for a safety net, particularly for underinsured children.

Committee Discussion

- Dr. Homer commented that he was unclear about the extent to which the medical health homes are currently in place and serving the needs of children who use the special health communities, especially those with heritable disorders, compared to other groups such as dual-eligibles. Ms. Comeau explained that nine states currently have health home SPAs. The ACA provision prohibits a health home SPA from concentrating on a specific population. Much of the focus has been on dual-eligibles and adults with chronic illnesses because they are the primary cost drivers. However, states can use other mechanisms, such as a provider network, to focus on specific populations. Rhode Island used this approach to expand the services provided to Medicaid-enrolled children with special health care needs. This highlights the importance of stakeholders working with their state

decision makers concerning the development of health home SPAs that address children's needs. Pediatric health home SPAs offer an opportunity to provide care coordination and family education and support as well as integrate mental health care with physical health care.

- Dr. Kus agreed that the focus has been on adults and on demonstrating that coordination of care reduces costs, hospitalizations, and emergency room visits. New York sees Section 2703 as a way to fund care coordination for children, including those identified through NBS. He indicated that he understood that the health home is a benefit that enhances the medical home and provides care coordination for children with more complicated cases. He hoped that this would prove successful in the Medicaid population and be adopted by other insurance programs. Ms. Comeau indicated that there is much fluidity in the way the terms "medical home" and "health home" are used. The terms are virtually interchangeable when used in the context of a philosophy of primary care. Under Section 2703, health homes are specifically defined and relate to benefits and funding support.
- Dr. Thompson, a subspecialist who handles children with complex chronic illness and coordinates the majority of their care, asked for clarification of the roles of primary care providers and specialists under the health home, especially with regard to reimbursements for services from the subspecialists. Ms. Comeau explained that subspecialists are eligible for reimbursement under Section 2703 SPAs. Integration between a variety of different kinds of providers is inherent in the design of the health home SPA.

General Discussion

- Mr. Suhr asked, via the chat function, how hospice care would be treated under the ACA. Ms. Comeau replied that there is nothing that delineates hospice care under Section 2703 SPAs. The ACA includes a provision that addresses hospice care for children with chronic conditions and life-threatening illnesses. Currently, children must have a life expectancy of six months or less and forego curative treatment in order to receive hospice care. This is problematic because children are physically dynamic and because some treatments can be both curative and palliative (e.g., radiation for brain cancer). The ACA removed the requirement of foregoing curative care and allows the use of concurrent care; the six-month life expectancy requirement remains in place. She directed the participants to the Catalyst Center's policy brief on financing pediatric palliative and hospice care that is available on the Center's website.
- Ms. Christelle Larose used the chat feature to ask whether there are caps on medical formula and low protein food. Ms. Comeau replied that these benefits have not changed in the states that mandate these services. Services are no longer limited in terms of dollar amounts, but benefits can be limited. This is a significant gap for families with children who have metabolic disorders, and there is nothing in the ACA that touches on this issue.
- Dr. Greene asked about the infrastructure for subspecialists who are not taking over primary medical home responsibility. Ms. Comeau believed that the enhanced Medicaid reimbursement would be allowed for subspecialists who are in coordination with a primary care practice. Each state Medicaid program will set this mechanism up differently. The Catalyst Center is developing a list of states with Section 2703 SPAs, and Ms. Comeau offered to help connect the Committee members with peer mentors concerning the process and promising practices.
- Dr. Greene asked for more information concerning the infrastructure that keeps these services available. Ms. Comeau indicated that the ACA includes some provisions concerning health information technology support. Because the Catalyst Center focused its work on the consumer protections, especially those related to children and families, she could not provide specific information on the technology provisions. She added that health information is a reimbursable service under Section 2703.
- Ms. Debbie Badawi indicated, via the chat function, that her state has considered allowing families on self-insured plans to buy a plan for their child(ren) with special health care needs on the exchange, if those plans are more comprehensive. She asked

whether this would be a reasonable option given that subsidies might not be available due to the availability of employer coverage. Ms. Comeau responded that the ACA exchange eligibility rules address the availability of minimal essential coverage (MEC). If an individual has access to MEC that is affordable, he or she is not eligible for the tax credits or subsidies that make purchasing coverage through the exchange affordable. One challenge is how to deal with children who have private insurance as their primary coverage and Medicaid as supplemental coverage. It is not clear whether these children will be able to switch from their private insurance to more affordable exchange coverage while keeping their supplemental Medicaid coverage. Currently, it does not seem as if this will be allowed, although there are ongoing efforts to make this possible.

V. Subcommittee Update – Current and Future Priorities and Projects

A. Subcommittee on Education and Training

Don Bailey, Ph.D., M.Ed.
Distinguished Fellow
Early Childhood Development
RTI International
Research Triangle Park, NC

Dr. Bailey reminded the participants that the overall charge of the Subcommittee is to review existing educational and training resources, identify gaps, and make recommendations aimed at parents, the public, and a variety of health professionals.

The Subcommittee has been working to identify one heritable condition, not on the RUSP and in which screening and treatment likely occurs later in child development, identify potential barriers for public health programs, and work with professional and parent organizations to identify the major education and training needs associated with the condition. Although the SACHDNC has focused on NBS, it is tasked with advising the HHS Secretary about both newborn and childhood screening. The Subcommittee chose to explore the issues and opportunities associated with other screening windows in order to support the process of developing recommendations through the use of exemplar conditions. In January, the Subcommittee selected three exemplar conditions on which it would base its work, and, recently, the Subcommittee learned of a similar effort that identified three exemplar conditions for later-in-life screening. The report on this effort has not yet been issued. The Subcommittee intends to review the draft report that is currently available for overlap and incorporate any lessons learned into the Subcommittee's own work. Dr. Bailey anticipated that the Subcommittee would seek input from the full Committee in September, gather input from stakeholders throughout the fall, and report back to the SACHDNC during the winter 2104 meeting.

The Subcommittee is also providing input on the 2013 Newborn Screening Awareness Campaign plans and activities, which include national meetings, an awards ceremony, a coffee table and e-book, and other outreach initiatives. The Subcommittee will also focus on ways to sustain the momentum of the campaign after the NBS fiftieth anniversary activities conclude later in the year.

The Subcommittee is also working to provide guidance to advocacy groups and others concerning the nomination and review process. To support this, the Subcommittee collaborated with the CRW to develop short, plain-language summaries of previously conducted evidence reviews and nominations that have not gone forward. This will help advocacy groups better prepare their nomination packages in the future. Ultimately, the Subcommittee anticipates improving the information available on the SACHDNC website and developing a book of case studies targeted

toward future nominators. Dr. Bailey anticipated that a draft document containing the summaries would be available for Committee review in the autumn.

B. Subcommittee on Laboratory Standards and Procedures

Kellie B. Kelm, Ph.D.
Scientific Reviewer/Biologist
Division of Chemistry and Toxicology Devices
Office of In Vitro Diagnostic Devices Evaluation and Safety
U.S. Food and Drug Administration
Silver Spring, MD

Dr. Kelm reported that the Subcommittee continued its work on the Clinical and Laboratory Standards Institute guideline IL-836, which addresses newborn bloodspot screening for Severe Combined Immunodeficiency (SCID). The Subcommittee received a presentation on the document in September 2012 and many members provided comments on it. The final document will be published soon.

One of the Subcommittee's priorities is providing guidance to state NBS programs for decision making concerning implementation, integration, follow-up, and quality assurance. Under this umbrella, the Subcommittee has been working on NBS case study definitions to harmonize NBS diagnoses for surveillance and epidemiology purposes. Currently, there are 150 records from 16 participating states. Definitions for metabolic and hemoglobinopathy cases are still in progress. Several states are pilot testing the case definitions as part of a project led by NewSTEPs. Once the pilot test is completed, the definitions will be provided to the Committee for review and recommendation.

A related project, also led by NewSTEPs, concerns the harmonization of quality indicators used by states to allow the comparison of data at a state level. Eight quality indicators have been targeted by the project for pilot testing. The ultimate goal is for states to use these indicators for data collection, starting when the NewSTEPs data base is up and running.

Another Subcommittee priority is the review of new and existing NBS technologies. The CDC and Dr. Matern have been working on a survey to determine the use of tyrosine versus succinylacetone (SUAC) to screen for tyrosinemia Type 1. The survey asked labs about their decision to use or not use SUAC screening and the obstacles to its adoption. The project began with 14 states and is now expanding nationwide. Once the survey is complete, CDC will analyze the data and present its findings.

Dr. Kelm also reported that a toolkit is being developed to help laboratories implement SCID screening.

C. Subcommittee on Follow-up and Treatment

Carol Greene, M.D.
Pediatric Genetics
University of Maryland Medical System
Society for Inherited Metabolic Disorders
Baltimore, MD 21201-1596

Dr. Greene reported on the current status of the Subcommittee project focused on developing a framework for assessing outcomes for NBS. The framework will help determine whether the anticipated benefits of NBS are being realized. The Subcommittee is working to identify the key

questions and the data types and sources needed to assess NBS outcomes as well as gaps that hinder the assessment, using sickle cell screening as an exemplar to test the framework. The framework includes several elements used to assess NBS outcomes and their related responses, measures, and data sources. Dr. Greene illustrated the use of the framework using the sample condition. The Subcommittee has been tracking work done by other groups, including those supported by HRSA, to ensure that this effort builds on their work and does not result in fragmentation. The Subcommittee will compare its framework with other existing standard data element sets and revise the framework as appropriate. The Subcommittee will also obtain feedback from stakeholders in the coming weeks. Dr. Greene anticipated presenting the revised framework and a draft paper on the background and rationale for it to the SACHDNC in September.

The paper on funding for and access to medical foods was recently published in Genetics in Medicine.

Forecasted activities for the Subcommittee include continued work on the integration of point-of-care screenings and bloodspot NBS, continued work on the framework for assessing NBS outcomes, and an exploration of the effects of the ACA on individuals and families with heritable conditions. The Subcommittee is particularly interested in identifying ways it can address questions of access to care as the ACA begins full implementation.

VI. Adjournment

Dr. Bocchini thanked the Committee members, the organizational representatives, and the public for their attention during the meeting and their contributions to the discussion. He also thanked HRSA for organizing the webinar. He indicated that Committee members would receive information on the May meeting in the near future. Ms. Sarkar thanked the participants for their thoughtful comments during the webinar.

Dr. Homer made a motion to adjourn the meeting, and Dr. Matern seconded the motion. Dr. Bocchini adjourned the meeting at 4:04 p.m.

Appendix A – Written Public Comments

Public Comment at April 19, 2013 SACHDNC meeting
Amber Salzman, PhD
President, The Stop ALD Foundation
amber@stopald.org
+1.610.659.1098

Hi, My name is Dr Amber Salzman and I lead the Stop ALD Foundation.
As a patient advocate I was very pleased to hear this morning that Secretary
Sebelius has put a discretionary committee in place while the Newborn Screening Saves Lives Act is up for reauthorization, thus enabling this committee to continue serving its critical role.

The purpose of my comments this afternoon is to provide an update on the newborn screen for Adrenoleukodystrophy (ALD), with the aspiration of moving the review process forward.

At the September 2012 SACHDNC meeting, the Adrenoleukodystrophy (ALD) newborn screening nomination was reviewed. The Committee recognized ALD "as a medically important disorder that deserves serious consideration, possessing a well-established case definition as well as screening, diagnostic, and treatment protocols." However, the Committee requested more prospective data from the Mayo Biochemical Genetics Laboratory (MBGL) pilot study prior to moving forward. Once additional data are available we were encouraged to contact the committee to facilitate an expedited review. The committee would then determine if data merits a formal review of the scientific evidence by the external condition review group.

With that context, I appreciate the opportunity to review the status of the MBGL pilot that is screening 100,000 Californian newborns:

- 50,000 samples have been screened and analyzed.
- The remaining 50,000 will be completed by the end of September 2013.
- Of the first 50,000 screened there were 12 that tested positive. After these
- 12 went through a second biochemical screen, only 6 samples tested positive.
- Of these 6 samples:
    - 3 came back negative for ALD from Molecular Test. Two of these samples were female and one was male.
    - 3 did not have sufficient material left to test, so CA testing lab is being asked to send additional material, and further testing will be done.
- In a blinded test 8/8 male newborn control samples came up positive.

Bottom Line:

- The ALD NBS test has a very low false positive rate 0.00024%
- The ALD NBS test correctly identified all positive control samples.
- Mechanisms are in place to do molecular screening on the samples that come up positive by biochemical screen.
- We do not want any more families to unnecessarily suffer the devastation
- ALD can cause when it is diagnosed too late to intervene.
- Given the recommendation from the September 2012 review of ALD, we hope you can provide guidance on how to best work with the SACHDNC to move forward expeditiously the review of ALD.

Also, as I'm sure the committee is aware, in March of this year NY voted to implement NBS for ALD. The legislation requires the NY NBS lab to validate the test and begin screening all newborns by January, 2014.

April 19, 2013

Public Comments … SACHDNC 30th meeting

Dear Committee and Sub-Group Members,

Greetings and thank you for your on going work on behalf of those with diseases detectable by a newborn screen. Your efforts, even though they are very tedious at times, are identifying sick children and saving lives. Thank you!

In these times of budgetary uncertainty and transitions please know that those of us out here are working to see that the Newborn Screen Saves Lives Act is renewed and that continuity is maintained in the work of the SACHDNC.

I have previously testified about my interest in having a renewed conversation about the potential of changing the criteria for committee approval of a newborn screen to not require a viable therapy. The implications of this are significant in terms of the philosophies, focus, work, and costs of social services, medical, and advocacy groups. Many organizations with disease communities similar to the MLD Foundation want to better understand the various considerations of knowing your child has a disease in advance of symptoms while you have time to prepare for the child's future, and then knowing the genetics to learn about how plan for any subsequent children. I would like to ask that anyone on the committee or in the audience that wants to join this conversation to please get in contact with me at Dean@MLDfoundation.org or via the other contact methods on the MLD Foundation website.

And finally, I briefly want to ask that the education committee consider creating, or making us aware if this is already available, materials that help us to work with families and organizations that are putting the NBS cart before the horse by forcing state legislation to implement screens well in advance of SACHDNC recommendations. I feel this legislation first approach is a train wreck waiting to happen that runs the risk of making it more difficult to implement even SACHDNC recommended screens in the future. Again, thank you for this time and for your ongoing work.

Sincerely,
Dean Suhr, President
MLD Foundation