

NEGC Update
February 27, 2009

ACHDGDNC Meeting: Notes from the Field

Hello: I am here in Washington DC at the Advisory Committee on Heritable Disorders and Genetic Diseases for Newborns and Children. It's actually been a nice break from writing the continuation application! There is so much happening at the national level right now, I thought it might be worth dropping a note to our NEGC constituents. As most of you know, this is a twice annual meeting which all Collaboratives are asked to attend. For those of you who do not attend regularly, this is a two day meeting which generally covers newborn screening and national policy. Most recently, the topic of Health Information Technology and national research, patient registries and long term follow up have been the focus of the committee and subcommittee meetings.

Today's agenda included subcommittee meetings (education, laboratory standards and long term follow up) and a review and (at times heated) discussion regarding the final draft report on the candidate nomination of Severe Combined Immune Deficiency (SCID).

The long term follow up subcommittee (chaired by Colleen Boyle) opened to a packed room—everyone wanted to be there as presenters included agencies which are perceived to have access to economic stimulus dollars. The goal of the committee meeting was to have attendees understand the purpose of LTFU data and data needs from the perspective of three different funding agencies—HRSA, NIH and the CDC. Michele Puryear clarified for the group that the three agencies come at this “from three different perspectives”; HRSA: service and communication process between pediatricians, specialty providers and NBS programs; NIH, research and CDC, surveillance.

After an IT presentation from the Office of the National Coordinator which covered “making the business case for HIT exchange and NBS”, representatives from the three agencies presented on their specific funded projects. These projects included 4 CDC funded NBS data initiatives (UT, CA, IA, NY). I have details of those projects if any of you would like them. Alan Hinman facilitated the discussion and summarized it best: How will we create the standardized data sets which, through the generation of new knowledge create the best possible outcome for NBS patient? He joked world peace would be easier to achieve and he might be right! The committee also heard presentations from across the country, including our own Janet Farrell who presented on the Massachusetts hearing screening early intervention program, outlining the Massachusetts data collection methods and links to early intervention. In addition, Sue Berry from Region 4 presented her point of service data registry and related regional projects (including Region I Priority Area 2), Mike Watson presented on the NICHD funded National Newborn Screening Translational Research Network and Cindy Hinton from the CDC presented on exploring areas of commonality in NBS data collection.

The ACHDGDNC convened in the afternoon to discuss the candidate disorder for NBS, SCID. Ellen Lipstein from Harvard presented the findings of the Evidence Review Workgroup. The final report on SCID were submitted January 2009, presented for discussion today. The Krabbe disease review is in progress. Dr. Lipstein began with a definition of SCID: a group of disorders characterized by absence of humoral and cellular immunity due to defects in T cell production and function. She stated the rationale for review

- Without disease specific treatment SCID leads to death early in childhood
- Earlier treatment (bone marrow transplant) may decrease mortality and morbidity

- Review includes: detailed methods and summarization of evidence
- Evidence: literature review (20 year time span) and an assessment of critical unpublished data from key investigators

The key findings of the committee included:

- Incidence of SCID is at least 1/100,000
- Population based trials are underway
- Some evidence shows early treatment may be more beneficial
- There is currently no systematic method of case finding, but pilot screening programs should serve to systematically id these cases
- Feasibility of screening: not clear, WI suggests screening is feasible
- Acceptability of screening: no evidence
- Treatment: critical evidence is lacking re: value of early treatment, cost effectiveness, adequacy of available treatment centers

The meeting was then open for public comment. Commentary, at times emotional, was submitted by Dr. Jennifer Puck, Dr. Mei Baker, Barbara Ballard (parent) and Marcia Boyle (advocate). The summary findings of the committee concluded there are strong reasons for screening including a “gold standard” diagnostic test, some indication that early diagnosis is important for treatment, and overall compelling treatment data. Honestly, there was strong division in the committee regarding whether all states should test at this time. The group agreed that there was demonstrated benefit for early identification of SCID including effective treatment, the fact that other conditions were uncovered during screening (e. De Georges Syndrome), identified children will not receive live vaccines, and that the family can avoid the “diagnostic odyssey”. However, there was some dissention including the issue that state programs are not ready to, conduct and pay for the test, the test has no quality control materials and as such, is not ready yet. In addition, there was philosophical disagreement about whether testing should be implemented when there are still pilot projects (MA and WI) in the field. One committee member made the point that a lab test can be highly accurate in a pilot test, but implementing a screening test for a condition which has an incidence of 1:100,000 is very different from a public health and training perspective. The group adjourned with the agreement that the committee would discuss at tomorrow’s meeting the elements needed to advance the SCID screen to a “Level A” recommendation.

That’s all for now. Again, I am happy to discuss details about any of the presentations. It has been a lively meeting so far, and it was certainly interesting watching the development of policy in action.

Regards, Amy

Amy Schwartz, MPH
 Assistant Clinical Professor & Associate Director
 NH Institute for Health Policy and Practice
 Project Manager, New England Genetics Collaborative
 Institute on Disability
 University of New Hampshire
 (603) 862-2962 (office)
 (603) 862-0555 (facsimile)
<http://www.negenetics.org>

