Archived Policy Statement

Heritable Disorders and Genetic Diseases in Newborns and Children

September 23, 2004
Sharon F. Terry

Thank you for the opportunity to make public comment today. Thank you also for the work of your committee—for the vision and leadership of the Genetic Services Branch of MCHB/HRSA and for the immense work of Mike Watson on the Uniform Screening Panel.

My name is Sharon Terry, and I am the president and CEO of the Genetic Alliance—a coalition of 600 advocacy organizations representing over 14 million individuals affected by genetic conditions, both common and rare.

I sit in this position not as a professional but because: I am the mother of two children affected by a genetic condition and the founder of a small disease-specific advocacy organization.

The advocacy community has a great number of concerns regarding newborn screening. My brief comments will be both general and specific.

A number of premises must be articulated when newborn screening is considered on the federal level. The first is overarching:

We are constrained by economic models developed within our crippled healthcare system—this conversation would be a different one in a nation with a more equitable alignment of resources.

Next, there are a series of basic premises for consumers:

- Parents want healthy babies—at any cost.
- Often parents do not know that:
- Their infants are being screened.
- Screening across states is variable.
- Attributes of tests such as sensitivity and specificity; analytical and clinical utility, and validity are variable.

- In lived experience, the odds of being affected are either 0% or 100%.
- Benefit analysis is not conducted from a medical model.

I would like to comment more fully on the last two points.
In lived experience, the odds of being affected are 0% or 100%.

In the moment that one receives a diagnosis, a line is crossed. One's worldview is quite different from the moment before the diagnosis. In the new experience, the discussion of odds—whether one or will or will not get a disease—becomes irrelevant and individuals have a poignant, though usually unconscious, understanding of public health perspectives verses personal health issues. The public generally assumes that odds apply to individuals. Consumers do not experience the test, diagnosis, and day-to-day struggles, on a population level—it is completely personal. The affected family, individual, or newborn uses its lived experience as the prism through which all life is assessed.

Consumers do not engage in benefit/harms analysis using a medical model, nor do they consider just the affected child in their decision-making.

In the minds of consumers, parents, decision-making about which tests should be part of a newborn screening panel is based on more than a narrow medical model. Families see benefit even in screening for conditions for which there is no treatment.

I recently asked our members about this issue. Here is an example of a reply from a mother who had two sons with Neimann-Pick disease that poignantly illustrates one reason why one might want to know about conditions for which there is no treatment—a criteria that in some systems is considered a hurdle.

After the death of her youngest son, she had her older son tested: *A year or two after Rick's diagnosis, when he was still apparently well, I asked him if he ever wished that he did not know that he had NPC, that he had never been tested. He said, "Oh no, Mom, now I know I am not stupid. I know there is a reason for some of the things I can't do." When we had thought that he had no obvious symptoms, Rick had been struggling to understand why he was not able to keep up with his peers, why there were some things that he could not do as well as he felt he should.*

Parents reported over and over to me that they need to know about genetic conditions in their family because they need to make informed decisions about lifestyle (for the family and the child), choosing caregivers and specialists, financial planning, choice of job, educational choice, finding a support group, securing insurance, aiding in building registries and participating in research.

Thus, the global context of decision-making and lived experience for parents includes more than what is traditionally considered in a medical model.
Now I turn to issues that are more specific:

Genetic Alliance has a Public Health Action Team-an active group of people concerned about newborn screening and other public health issues. Parents and professionals engage in daily discussion about many of the problems and potential solutions. I would like to share a brief synopsis of the more frequently-discussed concepts:

1.) It is a problem that there is inadequate understanding about NBS and the diseases associated with NBS. One part of the solution is the proactive work of parents and advocacy groups to raise the awareness of health professionals and the public.

2.) It is a problem that many communities lack necessary information and resources. Using the proposed uniform screening panel is a welcome recommendation, but the implementation goes beyond the tests. Parents and advocacy organizations stand ready to be part of the solution to deliver the services that must accompany more robust screening.

3.) It is a problem that technologies are advancing faster than policies, legislation, and treatments. Advocates have and will continue to promote effective public dialogue and decision-making..

4.) It is a problem that consistent, uniform, and continuous care is not available to babies, families, and Americans. The advocacy community initiates and sustains strong partnerships between parents, professionals, and the public.

Although your attention these two days is rightfully focused on the Uniform Screening Panel, I offer specific recommendations of the advocacy community for both the panel and the system. We request:

- **Proactive outreach to families and parents**
- **Input from underserved and underrepresented communities**
- **A uniform NBS panel of at least the recommended 30 tests, from state to state**
- **Resources for the Medical Home and for the necessary health professional education**
- **Health information accessible when and where it is needed**
- **National standards and INCREASED resources**

We ask this committee to recommend the Uniform Screening Panel, as determined by ACMG report, to the Secretary. We also ask that you recommend the Panel not as a stand-alone entity, but as a part of a larger comprehensive package that would include the above requests that would allow for:

- Resources to support the screening
- Mechanisms for collecting data after testing, post-market
- Systems that include resources beyond those usually included in the traditional medical model, including genetic counseling and services

Finally, we are aware that there are many “elephants” in the room—tensions between public and private labs, a lack of coordination among federal agencies, paternalistic and patronizing attitudes, and even “special interest and earmarking” behavior among advocates. I am, in the face of all these potential obstacles, reminded of what an advocate for NBS said in a recent email discussion about the Genetic Services Branch of MCHB: “For their leader, it is all about the babies.”

In the name of all of us who have crossed the “affected” line, I ask you to boldly, bravely, to make it “all about the babies.” Thank you.