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Heritable Disorders and Genetic Diseases in Newborns and Children

Comments to Secretary's Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children

May 5, 2005
by Sharon Terry

"We must ensure that all newborns have access to appropriate and effective screening programs"

Genetic Alliance—an international coalition comprised of more than 600 advocacy, research, and healthcare organizations that represent over 14 million individuals with genetic conditions and their interests—appreciates the opportunity to address this Committee regarding the Health Resources and Services Administration (HRSA) commissioned, American College of Medical Genetics (ACMG) report entitled *Newborn Screening: Toward a Uniform Screening Panel and System*. We would like to take this opportunity to commend both the Committee members for their dedication to improving the state of Newborn Screening in the United States, and HRSA for the leadership role they have taken on this issue.

As an organization representing millions of individuals living with genetic conditions, we know first-hand the vital role appropriate newborn screening programs play in the diagnosis and subsequent treatment of a disease. However, we are also painfully aware that the current system does not provide quality newborn screening services to the nation as a whole. As it stands, instead of a single standard that applies to all jurisdictions, individual states create and run their own newborn screening programs. Unfortunately, this lack of effective federal oversight, in addition to severe funding inadequacies, has resulted in dramatic variance in the breadth and quality of newborn screening programs across the country. Additionally, even today's best screening programs will eventually be unable to accommodate the rapid rate at which advancements in medical science and technology are made today. Allocating sufficient funds is the first—and perhaps the most important—step in improving our newborn screening programs. These programs desperately need funds to support testing, quality assurance, development of educational materials for professionals and consumers, and follow-up care.

That said, though funding is essential, it is certainly not the only barrier currently impeding the progress of our newborn screening programs. Therefore, in addition to supporting the uniform screening panel outlined by the HRSA commissioned ACMG report, **Genetic Alliance also recommends:**

- **A set of national minimum standards that would ensure uniformity in access to newborn screening services at all points of entry.**
- **Comprehensive educational programs—for both health care professionals and the public; equitable testing services; and thorough follow-up care.**
- **Quality assurance standards for all facets of any newborn screening program.**
- **An evaluation process for the maintenance and expansion of the core and secondary panels.**
- **Including a re-evaluation of the "efficacious treatment" and natural history criteria for inclusion.**

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The establishment of nationwide minimum standards will create uniformity of access in a system that is currently unbalanced.

The uniform condition panel outlined in the HRSA commissioned report will do a great deal to ensure that newborns in every state have access to adequate screening services. However, as the Committee is aware, the screening itself, while absolutely essential, is not nearly enough. Professional and consumer education about screening and appropriate follow-up care are both indispensable components of an effective newborn screening program, components that we know are just as vital as the laboratory tests themselves.

The document produced by the ACMG effectively outlines the tremendous benefits of creating a national newborn screening program that ensures that all babies born in the United States are screened for a minimum number of conditions, and it provides a tangible solution: the core and secondary screening panels. However, the existing variability in access to educational information will undoubtedly limit the effectiveness of these new requirements. While the Committee has acknowledged the need for new, more complete educational materials, Genetic Alliance would like to stress that any new materials produced must be multilingual, culturally sensitive, and appropriate for varying comprehension levels. The development of these materials must be a thoughtful process, engaging members of underserved and underrepresented communities. Similarly, even though scientists have discovered over 6000 rare diseases—some of which can be detected through newborn screening—health care providers do not always have the information or experience required to effectively manage those diseases in their patients, or even provide the appropriate referrals. Professional education, in addition to consumer education, must be a priority.

In addition, we must ensure that no newborn screening program ends the moment a test result—negative or positive—is delivered to the parent. Quality follow-up care is an indispensable component in any screening program, one that cannot be neglected or under-funded. When faced with a positive result, individuals and families must be provided with appropriate counseling, referrals, and services. Furthermore, in addition to easing what can be an enormous and unexpected burden on parents and families, follow-up programs should also benefit the scientific and medical communities. For example, screening programs that include a data collection component provide researchers with invaluable information regarding the natural history and treatment of a particular condition. Therefore, Genetic Alliance

recommends that the Committee establish guidelines for follow-up programs that include a data collection component.

In addition, though it has been said before, we cannot overemphasize the importance of providing resources—even those that are outside of the traditional medical model (e.g. support from disease advocacy groups, and other services)—for families as part of an appropriate and effective newborn screening program. Without fully integrating the educational and follow-up components, a newborn screening program cannot adequately serve practitioners and families. Additionally, though we are hopeful that a re-energized newborn screening program will ensure expeditious diagnosis and treatment of all children, we also recognize that any program created must prepare for the possibility of lapses in care. As such, we believe it is essential that the resources and services made available through newborn screening programs nationwide be accessible at all points of entry. In other words, a child who receives a diagnosis outside of the newborn screening system should have access to the same quality educational materials and follow-up care had by a child diagnosed through newborn screening.

A comprehensive newborn screening program must include quality assurance protocol that regulates testing and follow-up care.

In addition to discrepancies in the numbers of conditions for which state programs screen, there is a great deal of variance in the accuracy of the screening performed and the quality of the follow-up care provided to consumers around the country. As such, Genetic Alliance urges the committee to pay careful attention to the quality assurance portion of the ACMG document. A uniform screening panel does nothing to ensure balance in the national newborn screening programs if accuracy and quality standards are not consistent across all jurisdictions.

Since testing protocols (e.g. the cut-offs assigned to analyte levels) can vary a great deal between laboratories, a negative result in one state can be categorized as a positive result in another, or vice versa. This kind of inconsistency is both dangerous for babies and problematic for researchers examining the compiled data. Standards for the evaluation of laboratory tests and subsequent diagnoses, and guidelines for the retesting of samples, will help to ensure the kind of consistency that is essential for both treatment and research.

Furthermore, federal guidelines for newborn screening programs should address the current variance in turnaround time—the time between the birth of a baby and the delivery of the screening result to the family. Tests that are a part of a newborn screening program are, by definition, time sensitive; as such, we call for the establishment of a maximum turnaround time.

A review process ensuring the timely addition or removal of conditions to or from the core or secondary panel must be created and implemented.

If any newborn screening program is to remain effective over time, it is essential that its design be easily adaptable to future changes in the medical and scientific landscapes. As such, the absence of a clear-cut process for the evaluation of newly-developed or newly-established

tests in this report is an area of great concern for our organization and our member groups. When it comes to newborn screening, time really is of the essence; babies born today leave hospitals without receiving potentially life-saving screenings, a problem the uniform condition panel recommended by the ACMG addresses quite well. However, tomorrow, a new test may be developed or an older one invalidated. Without an expeditious evaluation process in place, the new tests cannot make it onto the core or secondary panel—or do not do so fast enough—and existing tests that are no longer reliable or necessary remain. In both cases, families suffer needlessly.

More specifically, as the ACMG's expert groups evaluated conditions for inclusion in the uniform panel, significant consideration was given to whether or not there was a "efficacious treatment" available. While Genetic Alliance respects the logic behind this particular qualification, we believe that the traditional medical model that this type of criterion reflects may not be the most appropriate one for newborn screening. That is, while the medical community may not consider a particular treatment "efficacious," an affected family might find that same treatment essential. Our community of consumers—14 million people living with genetic conditions—knows that the medical definition of treatment is more narrow and limited than the one they experience.

Furthermore, the ACMG report also required that diseases on the core panel have a "well understood natural history." Again, Genetic Alliance recognizes the basis for this stipulation, but we also know that many of the conditions for which there are—or will be—tests are extremely rare. For those conditions, a "well understood natural history" is a virtual impossibility, and will remain so unless we can (1) build registries and repositories of biological samples and clinical data through programs like newborn screening, and (2) track the progression of that condition over time. Newborn screening programs, if constructed and conducted properly, are public health initiatives that have the capability to positively impact both short-term and long-term health goals. Now is the time to rededicate ourselves to these programs and to health and welfare of this country.

Once again, I thank you for the opportunity to address this Committee. Should you desire it, I am happy to provide documentation to support these suggestions. Please, feel free to contact me.