



CLAIRE ALTMAN HEINE FOUNDATION, INC.

*for the prevention of Spinal Muscular Atrophy*

March 2, 2007

HFA-305  
Food and Drug Administration  
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Re: Docket #2006D-0336 and Docket #2006D-0347

To Whom It May Concern:

The Claire Altman Heine Foundation (CAHF) writes to express our concern with the IVDMA and ASR Draft Guidances proposed by FDA. We are a voluntary, not for profit organization dedicated to supporting pan-ethnic carrier screening for Spinal Muscular Atrophy (SMA) and educating the public and medical communities about SMA.

SMA is the leading genetic killer of children under the age of two. One in 6,000 babies is born with SMA and the majority of these children are stricken with the most severe form of the disease (Type I). These infants have a life-expectancy of just nine months and only five percent live to their second birthday.

SMA is relatively common; it is the second most frequent autosomal recessive genetic disorder after Cystic Fibrosis. About one in 40 individuals carries the diseased gene that causes SMA, which equates to over seven million Americans. This frequency cuts across all racial and gender barriers.

At the present time, there is no treatment or cure for SMA. The most effective means to combat the disease is through prevention in the form of pan-ethnic carrier screening. The technology to screen carriers for SMA has existed since 1996 in the form of a simple, accurate, and cost-effective blood test. However, there is little understanding or awareness of SMA among the OB/GYN or genetic counseling communities. Consequently, the test is not well utilized despite the fact that it can empower individuals of childbearing age to make more informed reproductive decisions.

As you know, SMA has a genetic cause, and like other such disorders, relies on accurate and accessible testing. We fear that the Draft Guidances as currently written may reduce the availability of these tests and slow innovation of future tests.

In diagnostic medicine, laboratory developed tests (LDTs) have paved the way for new and better tools to assess and treat a variety of diseases and disorders. With more than 1,000 LDTs currently in use, their importance cannot be overstated. Should FDA decide to implement either the



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IVDMIA Draft Guidance or the ASR Draft Guidance as currently worded, the vast majority of these LDTs may be banned or be stalled within a new legal and regulatory framework. For every person who could benefit from an early diagnosis for any condition, this delay would be unacceptable. In the oversight of laboratory testing, while safety and accuracy must be protected, so must the health and well-being of the patient. We believe there is a way to balance both of these priorities, and we encourage FDA to do so delicately, without overarching regulations that hold up essential progress.

Of additional concern, while existing LDTs wait for market approval under the new Draft Guidances, the production and innovation of new tests may be stunted. Diagnostic testing is extremely important for physicians, researchers, and patients alike, but the creation of new tests can be costly and time-intensive. If forced to work through an overly demanding approval process, it may prove difficult to devote efforts to innovating new tests, and testing for more uncommon conditions may become excessively expensive.

CAHF asks FDA to work toward a solution that keeps in mind both the needs of the laboratories and the patients they serve.

Thank you for your consideration,

Deborah Heine  
Executive Director