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Public Comment to Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children

Thank you for this opportunity and for your work on behalf of children and families with inborn errors of metabolism.

I am president and CEO of a start-up company, LysoPlex LLC, which is dedicated to developing technology discovered by Professor John Hopwood for screening newborns for lysosomal storage diseases. I come to the newborn-screening world from the perspective of the pharmaceutical and biotechnology industry where I have been involved with development of enzyme replacement and gene therapies for patients with lysosomal-storage diseases.

As you know, lysosomal-storage diseases or LSDs are a group of approximately 50 inborn errors of metabolism resulting from mutations in enzymes comprising the normal degradation pathway of cellular biomaterials. As a group, LSDs are among the most frequently observed genetic diseases with a combined incidence of 1 in 5,000. Put into perspective, the group of LSDs has an incidence approximately half that of cystic fibrosis but several times larger than many diseases currently screened.

The key difference with LSDs versus the classically screened diseases is that simple measures such as changes in diet, prophylactic penicillin or avoidance of fasting cannot improve the patient's outcome. Therapeutic action must be taken, and in many cases, be taken rapidly—before severe damage has taken place. Children with LSDs often experience severe mental retardation in many diseases or organ failure in others. The action needed involves replacement of the deficient enzyme through bone marrow transplant or exogenous replacement of the missing enzyme—enzyme replacement therapy. The Food and Drug Administration has recently approved a third approach: substrate inhibition therapy which helps in certain diseases. These measures are not inexpensive, nor in the case of bone marrow transplant, innocuous. In spite of their drawbacks, however, these therapies are the only option for these children and their families. In time, it is my hope that gene therapy or other curative technologies will be developed to definitively treat these children once they are diagnosed.

In every case, determining that the child has treatable LSD is the trigger that enables initiation of therapy. If treatment is sought after the beginning of symptoms, it may be too late. In some cases therapy must be initiated within days or weeks following the diagnosis whereas in other situations, it may be satisfactory to carefully watch the patient and initiate therapy at the first subtle signs of the diseases—well before critical organ function has been lost. Regardless of when therapy is initiated, we believe that providing the diagnosis to the parents can enable them to plan for the future; not only in establishing the prognosis of their current child but also in planning for future pregnancies. One of the tragedies we hope to avoid is the subsequent birth of affected children while the parents are still trying to gain a diagnosis for the first child. This happens all too frequently.

So, what requests does LysoPlex have of this committee and the Federal Government?

1. Continued advocacy for universal access to newborn screening

We acknowledge that States have primacy in the conduct of newborn screening. However, all children born in the United States should be protected through universal and equal access to testing—regardless of where the child is born. The current system is complex and unwieldy, particularly when it comes to payment for newborn screening. The marked variability of diseases tested among the States is a disappointment. Concentration of expertise in regional centers with substantial Federal support could be a route to universal testing.

2. Continued support for development of new technology which enables testing for inborn errors of metabolism beyond the current group of screened diseases

LysoPlex is taking forward a novel multiplex technology which enables simultaneous measurement of multiple lysosomal proteins. As small, flat organizations, biotechnology companies often can translate discoveries into approved products faster than other organizations—especially when combined with ready access to the knowledge and resources of the Federal government. The ability to rapidly mobilize these Federal resources could be extremely beneficial to start-ups that must often bootstrap themselves from a very modest base prior to professional investment.

3. Continued leadership role in the establishment of national recommendations which will encourage States to rapidly incorporate these new technologies into state screening procedures

The work conducted by this committee in defining what diseases should be screened for is crucial. In addition we propose that the Federal Government also provide the financial backing and incentives for states to broadly incorporate these national recommendations into their standard newborn screening procedures.

Once again, thank you for your time and your service to these children.

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