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Federal Advisory Committee Recommends Pompe Disease for Newborn Screening

WASHINGTON – May 17 2013- In a long awaited meeting, the Secretary's Discretionary Advisory Committee for Heritable Disorders in Newborns and Children (DACHDNC), in a vote of 11 – 2, recommended the addition of Pompe Disease to the recommended uniform newborn screening panel (RUSP). This recommendation will be sent to the Secretary of Health and Human Services to approve adding Pompe to the RUSP. The letter to the Secretary will highlight the need for states currently implementing Pompe screening to coordinate activities and help establish a framework for state-based screening. The committee considered both the significant benefit to screening for infantile as well as the need for more studies about the optimal management of those with late onset.

"Genetic Alliance applauds the thoughtfulness of the committee in deliberating this important topic" state Natasha F. Bonhomme, vice president of strategic development and director of maternal and child health at Genetic Alliance. She adds, "there is still much to discuss about what population based screening for Pompe will entail and we can best support states through the implementation process, but this vote is a forward step." Much of the public discussion (via chat feature on the webinar) centered on the inclusion of cost and screening impact on state newborn screening programs in the discussions regarding recommending Pompe.

Pompe disease is an inherited condition that affects many different parts of the body. It is a lysosomal storage disorder because people with Pompe disease have lysosomes (the recycling center of each cell) that cannot break down certain types of complex sugars. This causes undigested sugar molecules and other harmful substances to build up in cells throughout the body, resulting in a variety of symptoms. Pompe disease has a broad spectrum of illness. The infantile is the most severe, with children showing symptoms before 12 months of age. Without treatment, death occurred in early childhood. For those with the late onset disease, adults typically receive a diagnose up to 10 years after first seeking care for symptoms.

Screening is based on measuring enzyme activity and genotyping is used to diagnosis the condition as well as identify carriers, predicts infantile versus late onset disease. Treatment for Pompe disease is enzyme replacement therapy (ERT). There are two drugs currently available, both produced by Genzyme. The treatment protocol includes infusion every two weeks.

Missouri began screening for Pompe on January 15, 2013. In the first four months of this screening, 1 case of classic infantile-onset was found with 7 other cases needing further examination. Though most groups note that prevalence is 1 in 40,000, data presented today during the DACHDNC meeting indicate the prevalence maybe as high as 1 in 28,000 based off of a recent study done by University of Washington using anonymous dried-blood spot study.

One of the biggest concerns regarding implementing population screening for Pompe is that majority of people identified though screening would have the late onset variation of the condition, meaning they would not be symptomatic until after childhood. Screening for conditions that impact children early in life is a cornerstone of newborn screening. Alex Kemper, MD, MPH, MS, who gave the report from the Condition Review Workgroup, mentioned that there are processes for follow up for those identified with late onset.

The DACHDNC was established earlier this year to fulfill the role that previously was filled by the Secretary's Advisory Committee on heritable Disorders in Newborns and Children (SACHDNC). The SACHDNC was originally chartered in February 2003 to advise the Secretary of the U.S. Department of Health and Human Services regarding the most appropriate application of universal newborn screening tests, technologies, policies, guidelines and standards for effectively reducing morbidity and mortality in newborns and children having, or at risk for, heritable disorders. The Newborn Screening Saves Lives Act of 2008 further defined the SACHDNC and included

the provision that the Secretary of Health and Human Services must respond to recommendation within 180 days. The charter of SACHDNC expired in April 2013 and will be renewed when the Newborn Screening Saves Lives Reauthorization Act passes Congress. Advocacy groups hope this legislation will pass by the end of they year.

The SACHDNC adopted a list of 29 recommended conditions in September 2005. Since that time, based on the recommendations of the SACHDNC, the Secretary of Health and Human Services has added Severe Combined Immunideficiency (SCID) and Critical Congenital Heat Disease (CCHD) to the RUSP for inclusion into mandatory newborn screening conducted by state public health programs. As of today, no state screens for all conditions on the RUSP, though many states screen for 29 out of the 31 conditions recommended by the Secretary of Health and Human Services.

To date, twelve conditions have been brought to the Advisory Committee: 22q11.2 Deletion Syndrome, Adrenoleukodystrophy, Critical Congenital Heart Disease, Fabry Disease, Hemoglobin H, Krabbe Disease, MPS 1 (alpha-L-idurinidase deficiency), Neonatal Hyperbilirubinemia, Niemann-Pick Disease, Pompe Disease, Severe Combined Immunodeficiency (SCID), and Spinal Muscular Atrophy (SMA). Thus far, only SCID and CCHD have been officially added to the uniform screening panel. There is currently a formal process for individuals or organizations to nominate a heritable disorder to be considered for inclusion in the recommended uniform screening panel (http://www.hrsa.gov/heritabledisorderscommittee/nominate.htm).

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