

Ms. Barbara Vorpahl
Chair
National Niemann-Pick Disease Foundation
P.O. Box 49
401 Madison Avenue, Suite B
Ft. Atkinson, WI 53538

Dear Ms. Vorpahl:

The Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) received your nomination form and all accompanying materials (nomination package) for Acid Sphingomyelinase Deficiency/Niemann-Pick Disease, types A and B on May 2, 2008. The ACHDNC reviewed the nomination package and reported the results of that review on July 25, 2008. Based on a review of the nomination form and accompanying materials, the decision was made not to send the nomination package forward to the ACHDNC's evidence review group at this time.

Although Acid Sphingomyelinase Deficiency/Niemann-Pick Disease, types A and B, is medically serious and the spectrum of the disorder is well described, several factors limit its appropriateness for inclusion in newborn screening at this time: a) no population-based data is available, although some clinical studies derived from major referral centers are reported; b) discerning type A from B is not always possible to help predict the phenotypic range of those children who will be identified through population-based screening; c) characteristics of the screening test(s) are unknown since no population-based assessments have been performed; d) no published studies are available to show the efficacy of treatment in humans especially for those most likely to benefit early in life, although clinical trials are underway; e) no FDA approved treatment currently exists; and f) the need for pilot studies of the newborn screening test and treatment protocols.

The ACHDNC is available for further discussion of screening newborns for Acid Sphingomyelinase Deficiency/Niemann-Pick Disease, types A and B. You are welcomed to resubmit a nomination package when there is sufficient evidence to address the issues above. The next ACHDNC meeting is February 26-27, 2009. ACHDNC may be contacted at (301) 443-1080 if you wish to make public comments at this meeting.

Thank you for your interest and support of newborn screening programs and the ACHDNC's activities.

Sincerely yours,

/s/

R. Rodney Howell, M.D.
Chairperson