Nebraska Department of Health and Human Services
Division of Public Health
State of Nebraska
Dave Heineman, Governor

To:

From: Jeff Hoffman, MS, CCC-A, Early Hearing Detection & Intervention Program Manager
Nebraska Dept. of Health & Human Services, Division of Public Health

Re: Test for genetic diagnosis of hearing loss for
Date of Birth

According to our records has been confirmed with permanent childhood hearing loss. This letter is to remind you of the availability of genetic testing to help with the further diagnosis and treatment of this infant. If you request retrieval of the residual dried blood spot (DBS) from this infant's newborn screen by 90 days of age, it can be returned to you and may be submitted for testing for such causes of hearing loss as congenitally-acquired cytomegalovirus (CMV), Connexin 26, Connexin 30, Mitochondrial mutations or Pendred syndrome. (Note: The DBS are destroyed after 90 days).

- Congenital CMV is the most cause of 30-40% of congenital and later-onset permanent hearing loss. The newborn dried blood spot is the best way to determine if CMV is congenital or acquired later.
- Connexin 26 is the most common cause of non-syndromic genetic deafness, responsible for about 50%.
- Diagnosis of syndromic hearing loss can be especially useful for management. Some common causes of syndromic hearing loss for which DNA testing is available are mitochondrial conditions and Pendred syndrome.

If you choose to request retrieval of this infant's dried blood spot (DBS), please complete all of the following steps:

1. Call Julie Miller, Newborn Screening & Genetics, at 402-471-6733 to request that the laboratory hold the DBS specimen until you submit the enclosed Requisition.
2. Have the parent/guardian complete and sign the first section of the Requisition.
3. Fill in your name and address, sign and date the bottom section of the Requisition.
4. Fax the Requisition to PerkinElmer Genetics, Attention: Bob Wise at 412-220-0784.
5. Call PerkinElmer Genetics at 412-220-2300, ext 120 to confirm the receipt of the fax.
6. Mail the original Requisition to: PerkinElmer Genetics
   90 Emerson Lane, P.O. Box 219
   Bridgeville, PA 15017