Secretary's Advisory Committee on Heritable Disorders in Newborns and Children
5600 Fishers Lane, Room 18A19
Rockville, Maryland 20857
(301) 443-1080 – Phone
(301) 480-1312 – Fax
http://www.hrsa.gov/heritabledisorderscommittee

May 9, 2012

John Routes, M.D.
Chief, Section of Allergy and Clinical Immunology
Professor of Pediatrics, Medicine, Microbiology and Molecular Genetics
Department of Pediatrics
Medical College of Wisconsin
9000 W. Wisconsin Ave.
Milwaukee, WI 53226-4874

Dear Dr. Routes:

The Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) received your nomination form and all accompanying materials (nomination package) for 22q11.2 deletion syndrome (22q11.2 DS) on July 20, 2011. The SACHDNC reviewed the nomination package and reported the results of that review on January 26, 2012. Based on a review of the nomination form and accompanying materials, the SACHDNC decided not to send the nomination package forward to the SACHDNC's external evidence review group at this time.

The 22q11.2 DS is medically serious and the spectrum of the disorder is well described. However, the SACHDNC decision was based upon the fact that the requirements have not been met to forward 22q11.2 DS to evidence review and subsequent consideration for inclusion in the Recommended Universal Newborn Screening Panel (RUSP).

Additional data are needed in the following areas:

a) The proposed test has been studied in major referral centers in patients with congenital heart disease known to have 22q11.2 DS; however, no population-based data is available.
b) Performance characteristics of the multiplex, real-time PCR (Polymerase Chain Reaction) screening test, for detection of TBX1 haploinsufficiency with confirmatory testing with FISH or high resolution microarray analysis, have not been assessed in a population-based study.

c) Without population-based data, the impact of testing on diagnosis is unclear.
d) No published studies are available to show the benefits or effectiveness of early treatment in mild cases, or in cases without life-threatening manifestations, if diagnosed early in life. Thus, there is a need for pilot studies of the proposed newborn screening test algorithm and treatment protocols to provide the data necessary to inform an evidence review.

The SACHDNC is available for further discussion of screening newborns for 22q11.2 DS. You are welcome to resubmit a nomination package when there is sufficient evidence to address the issues listed above. The next SACHDNC meeting is May 17–18, 2012. Please call (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 SACHDNC's activities.

Sincerely,

/s/

Joseph A. Bocchini Jr., MD
Chairperson