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## APHL Position/Policy Statement

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### Newborn Screening Follow-up

#### A. Statement of Position

The Association of Public Health Laboratories (APHL) recognizes that infant tracking and follow-up of presumptive positive cases and unsatisfactory specimens are critical parts of the newborn screening system and must be integral to any program considerations concerning testing, financing and parent/provider education. Further, APHL endorses the follow-up considerations previously published by the American Academy of Pediatrics Newborn Screening Task Force (1), and supports the two published reports of the Council of Regional Networks for Genetic Services regarding newborn screening systems (2,3). Successful follow-up ensures that newborn screening provides the effective interventions necessary for improved outcomes for all conditions included in the state's test panel.

#### B. Background Supporting Position

Newborn screening is a multi-faceted system of newborn preventive care (1-4). Typically, newborn screening programs are organized such that there are two major areas of result follow-up: specimens are unsatisfactory for definitive analysis, and specimen results are outside of the expected range. Follow-up must be performed using a written protocol coordinated throughout the entire newborn screening system. The newborn screening system must ensure that follow-up activities accomplish the goal of locating the infants in time to avert catastrophic consequences.

Short-term follow-up refers to the process of ensuring that all newborns are screened, that an appropriate follow-up caregiver is informed of results, that confirmatory testing has been completed, and that the infant has received a diagnosis and, if necessary, treatment. Written protocols defining specific activities, time lines, and the beginning and ending of responsibilities for each component of the screening and follow-up system are essential, and are typically based on defined and assigned responsibilities.

There must be a quality assurance mechanism within the follow-up system for the reporting of final diagnosis and pertinent clinical information, for quality care of patients during follow-up and beyond, and for monitoring the appropriate databases linkage and information exchange throughout the United States. A system for sharing information among all newborn screening programs should be routinely available. This sharing is essential to minimize or eliminate the unnecessary duplication and multiplication of databases.

Program financing must address the actual cost of follow-up including not only communications of results, but also assurance that appropriate confirmatory testing and treatment have been implemented and that necessary services, including satisfactory medical home, are in place. Infant tracking for presumptive case follow-up may occur either as a laboratory function or as a separate but integrated system function, and should adhere to a written protocol. This protocol should define and assure appropriate diagnostic evaluation including confirmatory laboratory testing, case definitions, medical management standards, and linkages to a suitable medical home, as well as feedback to the laboratory concerning case outcomes for quality control and quality assurance purposes. Mechanisms for tracking,

follow-up and resolution of unsatisfactory specimens and inconclusive test results must also be included in the overall follow-up system and routinely evaluated to assure tracking effectiveness. Newborn screening programs should attempt to collect and monitor long-term outcomes for infants with positive screening results and confirmed disease. The extent of such efforts will be dependent on available infrastructure and program resources. Because long-term outcomes could involve issues surrounding the retention and use of the original screening specimen, newborn screening systems should include a policy defining protocols for specimen storage, consent and access for future use.

### **C. References**

1. American Academy of Pediatrics Newborn Screening Task Force. Serving the family from birth to the medical home: a report from the Newborn Screening Task force convened in Washington DC, May 10-11, 1999. *Pediatrics* (suppl) 2000;106: 423-427.
2. Therrell BL, Panny SR, Davidson A, et al: U.S. Newborn Screening Program guidelines. Statement of the Council of Regional Networks for Genetic Services, Screening 1992.1:135-47.
3. Pass KA, Lane PA, Femhoff PM, et al. U.S. Newborn Screening Program Guidelines for follow-up of children, diagnosis, management and evaluation. Statement of the Council of Regional Networks for Genetic Services (CORN). *J Peds* (supp Q 2000; 137:144)
4. Hannon WH, Boyle J, Davin B, Marsden A, McCabe ERB, Schwartz M, Scholl G, Therrell BL Jr, Wolfson M, Yoder F. Blood collection on filter paper for neonatal screening programs; approved standard—Third edition. NCCLS document, LA4-A3. NCCLS, Wayne (PA) 1997;17:1-23.

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