



# APHL Position Statement

## Newborn Screening Follow-up

### A. Statement of Position

The Association of Public Health Laboratories (APHL) acknowledges and endorses the follow-up considerations previously published by the American Academy of Pediatrics Newborn Screening Task Force, the Council of Regional Networks for Genetic Services regarding newborn screening systems, and Clinical and Laboratory Standard Institute.<sup>1-4</sup> Further, we support the use of the Performance Evaluation and Assessment Scheme (PEAS) developed by the National Newborn Screening and Genetics Resource Center in assessing and improving all aspects of the newborn screening system.<sup>5</sup> We also acknowledge and support the clarifying definition of long term follow-up published by the Secretary's Advisory Committee on Heritable Diseases in Newborns and Children that includes ensuring effective interventions for improved outcomes for all patients with screened conditions.<sup>6</sup> APHL takes the position that tracking and follow-up of out-of-range newborn screening laboratory test results and invalid specimens are critical parts of the newborn screening (NBS) system, and the screening program must work to include these activities in planning, financing, implementing, and evaluating pre-analytic, analytic and post-analytic NBS system activities.

### B. Background/Data Supporting Position

NBS laboratory operations in some state public health programs. Where it is not a laboratory

function, the laboratory plays an important supporting role in providing timely and accurate data. Typically, public health NBS 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 of results for normal newborns. Follow-up should be performed using written protocols that coordinate follow-up care so that newborns can obtain confirmatory testing and diagnosis in time to avert catastrophic consequences. In addition to providing case definitions, these protocols should define and seek to assure appropriate diagnostic evaluation (including confirmatory laboratory testing), medical management, and connection to a suitable medical home.

Short-term follow-up refers to the process of ensuring that all newborns are screened, that an appropriate caregiver is informed of results, that repeat testing on a new specimen or 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 should be based on known screening windows (defined as the time between when the abnormal analyte is detectable by NBS and the time at which adverse consequences can occur). Written

protocols should also address at what point a case is considered lost-to-follow-up after exhausting all reasonable efforts as defined by protocols. Long-term follow-up begins at the time of diagnosis and extends throughout the life of the diagnosed individual.<sup>4</sup>

A quality assurance mechanism should exist in short-term follow-up that ensures a final diagnosis, supporting laboratory results, and pertinent clinical information at the public health program level. Long-term systems of care should include data collection and analysis using program-defined indicators to monitor anticipated health outcomes. Program changes based on quality assurance data should be made to facilitate optimum health outcomes. As an aid in information exchange across programs, the use of comparable measures nationally is encouraged. In summary, short-term case finding data should be reported in a timely way to the National Newborn Screening Information System.<sup>7</sup>

### 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* (supplement) 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; I37:144).
4. Tuerck J, Dhondt J-L, King P, et al. Clinical Laboratory Standards Institute. Newborn screening follow-up; Approved Guideline. CLSI document I/LA27-A. Wayne, PA: CLSI, 2006.
5. Therrell BL, Schwartz M, Southard C, et al. Newborn

Screening Performance Evaluation Assessment Scheme (PEAS). *Sem Perinatol*. 2010; 34(2):105-20.

6. Kemper AR, Boyle CA, Aceves J, et al: Long-term follow-up after diagnosis resulting from newborn screening: statement of the US Secretary of Health and Human Services' Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children. *Genet Med*. 2008; 10(4):259-61.
7. Therrell BL, Hannon WH. National evaluation of U.S. newborn screening system components. *Ment Retard and Develop Disabil Res Rev* 2006; 12:236-45.

### D. Other References of Interest

- A. Hoff T. Long-term follow-up of culture in state newborn screening programs. *Genet Med* 2008 Jun; 10(6):396-403.
- B. Hoff T, Ayoob M, Therrell B. Long-term follow-up data collection and use in state newborn screening programs. *Arch Pediatr Adolesc Med* 2007; 161:994-1000.
- C. Hoff T, Hoyt A, Therrell B, Ayoob M. Exploring barriers to long-term follow-up in newborn screening programs. *Genetics in Medicine* 2006; 8:563-70.
- D. Hoff T, Hoyt A. Practices and perceptions of long-term follow-up among state newborn screening programs. *Pediatrics* 2006 Jun; 117(6):1922-9.

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