CLSI NBS Follow-up Guidelines
Sub-Committee

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“FOLLOW-UP”

The verb: To maintain contact to evaluate a diagnosis or to determine the effectiveness of treatment; to take appropriate action. …Webster

The noun: The people doing follow-up
Follow-up Personnel Responsibilities

- Follow-up
- Education
- Administration
Newborn Screening Follow-up

**Short-term**: birth to diagnosis

**Long-term**: diagnosis throughout life
Essential Follow-up Functions

- All “abnormals” are followed to diagnosis and assurance of intervention (short-term FU)
- All other FU referrals are resolved
- Every eligible newborn has a valid screening result
- Collection of long-term FU data for program evaluation
Follow-up Personnel Need:

- Knowledge of conditions
- Knowledge of confirmatory services and how to access them
- Intimate knowledge of the birth facilities and practitioners within the screening jurisdiction
- A network of community services to assist, i.e., public health, law enforcement, SCSHCN, treatment centers, etc.
- A person who is tenacious, resourceful, not easily frustrated, persuasive, tactful, etc.
Types of Follow-up

PASSIVE: A report is sent to the submitter, with no further action on the part of the nbs program. (normal, carrier info, early testing)

ACTIVE: Ensures that appropriate actions are taken to resolve cases within specified time frames (abnormal, inadequate)
Categories of Follow-up

- Abnormal results
- Unsatisfactory screening
  - Not done
  - Inadequate
  - Too early
- Carrier and Risk factor
Follow-up Load*

- Abnormal .............. 1.5% (60,778)
- Inadequate screen... 2% (0.06-11%)
- Too early.............. 17% <24 hours
- Not done............... ~1%
- Carrier and risk factor.............. Unknown (5-10%)

*NNSGRC, National NBS report-2000
Follow-up Goals

**Emergent disorders**: on tx by 10 days (galactosemia, CAH, organic acidemias, urea cycle defects, fatty acid oxidation)

**Non-emergent disorders**: on tx by 3 weeks (PKU, CH, biotinidase, sickle cell disease)

**Hearing Loss**: EI by six months
Days to Treatment

Specimen Collected: 1-7+ days
Transit Time: 1-10+ days
Screening Lab: 1-5+ days
Follow-up: 1-30+ days

AGE AT DX: 4-51+ DAYS
Days to Treatment: Emergent Conditions*

1990:
- <10 days: 30%
- >10 days: 30%
- unknown: 40%

2000:
- <10 days: 50%
- >10 days: 20%
- unknown: 30%

Goal:
- <10 days: 80%
- >10 days: 20%
- unknown: 0%

*NNSGRC, National NBS report-2000
Days to Treatment: Non-emergent Conditions*

* NNSGRC, National NBS report-2000
Infants Lost to Follow-up 2000*

- Abnormal: 60,788
- Lost to follow-up: 1,609 (2.6%)
- Deaths: 45 (21 deaths involved abnormal results for CAH, Gal, MSUD)

*National NBS Report, 2000
Problems in Follow-up

- Varies widely in quantity and quality
- Most are not measuring their own activities, but instead program goals
- Statistics support poor performance in meeting dx goals
- FU priorities may not be clear
Problems in Follow-up

Follow-up coordinators don’t have the time or the expertise to devise FU studies

Coordinators may have difficulty advocating for themselves within the screening system and political milieu

No guidelines for FU

No standard for FU educational qualifications (RN’s, GC’s, secretaries)
Follow-up: The Last Frontier

“Active” FU programs began in the 70’s and 80’s

All U.S. screening programs have FU personnel and procedures, however:
- No survey of follow-up practices has ever been done
- Efficacy of any given FU procedure is unknown
- No published studies on the effectiveness of FU activities within a screening system
Follow-up: The Last Frontier

- Last portion of the NBS system to develop guidelines
- FU folks have struggled for equal status within the screening system; ie we are not represented on this Advisory Committee
- FU activities are often under funded, although this is changing thanks to HRSA and CDC
Intent of Guidelines

To provide a framework and best practices model to ensure timely identification of affected infants
Exclusions/Limitations

Analytical portions of the screening system and/or confirmatory testing

Treatment Modalities
Intended Audience

Global document applies to those involved in any aspect of nbs follow-up:

- NBS follow-up personnel
- Maternity and newborn health care providers
- Medical home provider
- Confirmatory services/sub-specialty providers
- Parents
Over Arching Principles

- FU is an integral part of the nbs system
- FU should be centralized
- FU activities should be uniform across conditions, jurisdictions....
- FU activities should be prioritized
- FU should be active for abnormal and inadequate cases
- FU should be accomplished quickly
- All cases should be resolved
- FU activities need evaluation
FU Guidelines

- Define FU and its place and function within the system
- Outline FU responsibilities
- Describe the communication and data systems essential to FU
- Policies and Procedures of FU
- Quality assurance and evaluation
- Outline research needs
Research Needs in FU

- Survey of policies and procedures
- Efficacy of FU policies and procedures
- Costs of FU by FU category
- Evaluation of lost to FU cases and how they get lost
- Evaluation of fact sheets on provider knowledge and performance
- Impact of MS/MS on FU
- Impact of carrier detection on parents/newborns and FU
CLSI Timeline

**September, 2004:** Subcommittee meeting

**May, 2005:** Subcommittee vote on draft

**June, 2005:** Area Committee vote

**August, 2005:** Proposed document review and comment by CLSI delegates, board; public review

**February, 2006:** Revisions complete

**Feb-May, 2006:** SC, AC, Delegate and Board votes

**June, 2006:** Publish Approved Guidelines