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 fu, ie 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
  ..... 50% <48 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

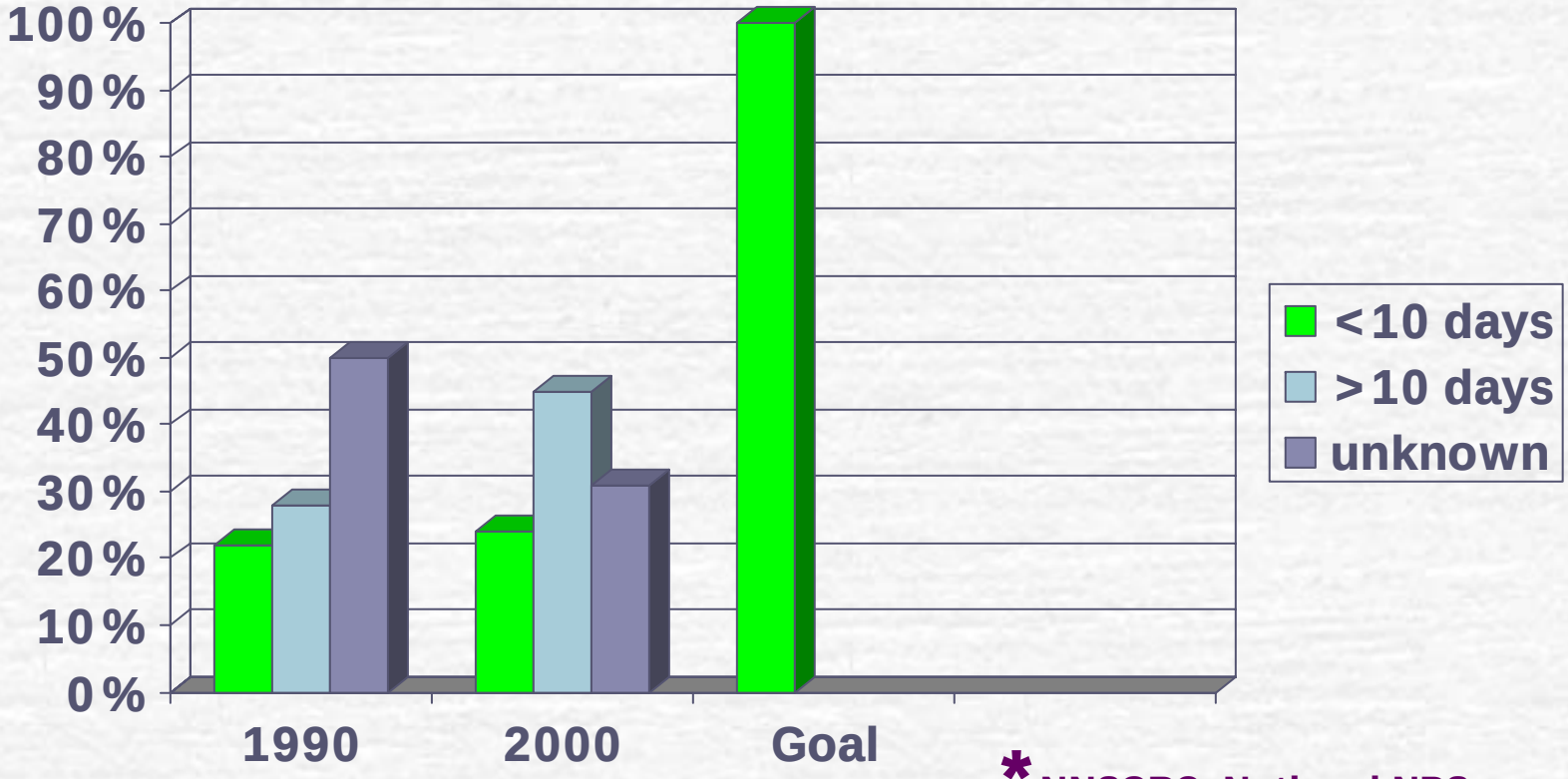
Specimen Collected:	**1-7+ days**
Transit Time:	**1-10+ days**
Screening Lab:	**1-5+ days**
Follow-up:	**1-30+ days**

AGE AT DX: 4-51+ DAYS

Days to Treatment: Emergent Conditions*

* 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

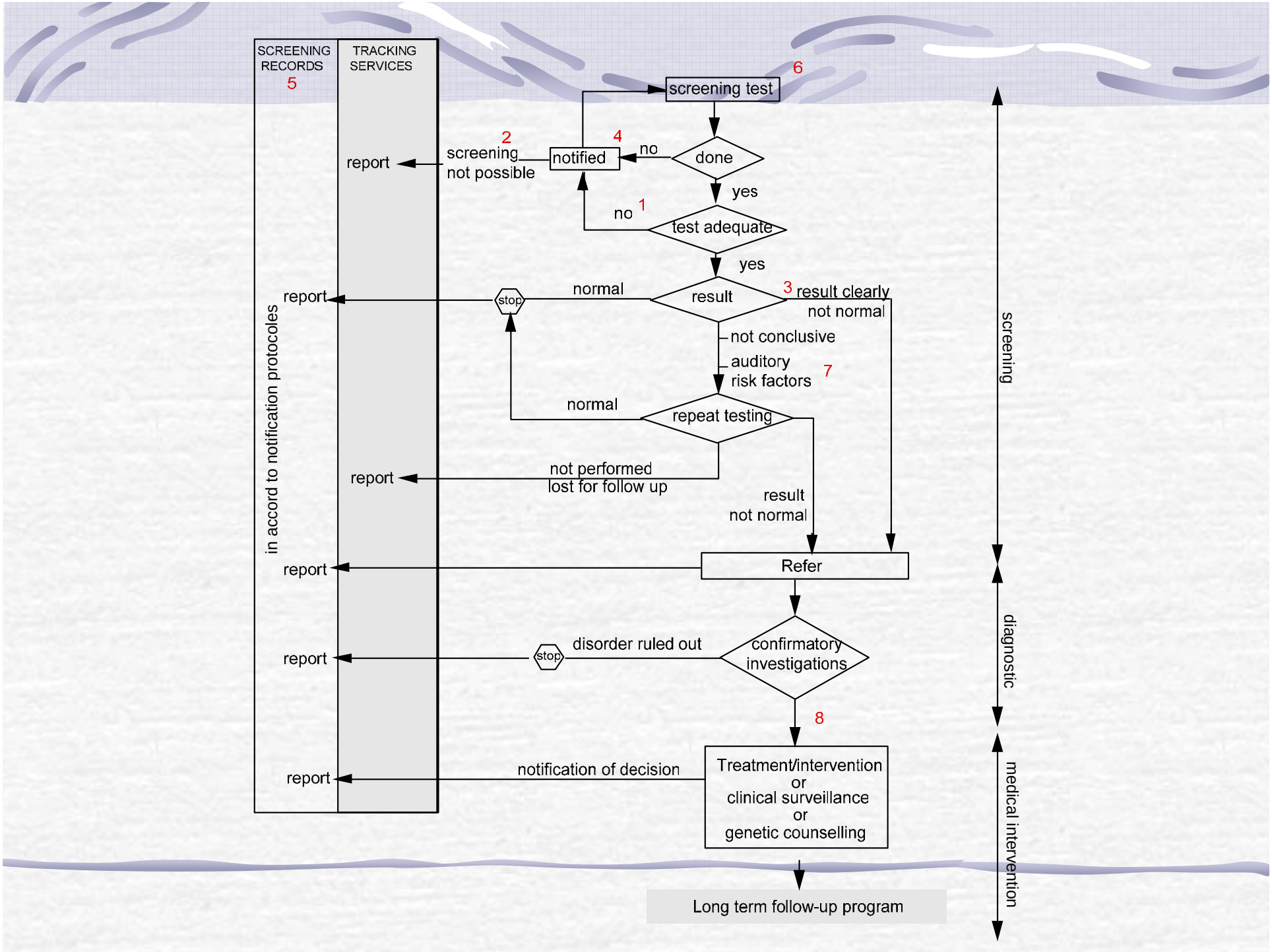
Exclusions/Limitations

- 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


SCREENING RECORDS
5
TRACKING SERVICES
in accord to notification protocoles
6
screening test
2
screening not possible
report
4
notified
no
done
yes
no
1
test adequate
yes
report
stop
normal
result
3
result clearly not normal
not conclusive
auditory risk factors
7
normal
repeat testing
report
not performed lost for follow up
result not normal
report
Refer
report
stop
disorder ruled out
confirmatory investigations
8
report
notification of decision
Treatment/intervention or clinical surveillance or genetic counselling
Long term follow-up program
screening
diagnostic
medical intervention

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