

Attachment 6
Case Report Form

Case Report Form

1. State or military base in which the birth occurred _____

2. Condition (must have been screened for in state of birth at the time of birth)

(Check one)

- Classical phenylketonuria
- Primary congenital hypothyroidism
- Galactosemia (any type if law is not restrictive)
- Maple syrup urine disease
- Homocystinuria
- Biotinidase deficiency
- Classical congenital adrenal hyperplasia (saltwasting or simple virilizing)

- Sickle cell disease

3. Child's birth month/year _____

4. Reason(s) for missed or delayed diagnosis (choose all that apply):

Specimen collection

- No specimen collected
- Specimen mislabeled
- Transfer to another hospital, no specimen collected
- Delayed first specimen because of home delivery
- Delayed first specimen because infant was in NICU
- No satisfactory specimen collected (initial specimen invalid and repeat not obtained)
- First specimen was unsatisfactory, repeat obtained after delay
- Other (explain)

Additional details if available:

Specimen transport

- Specimen not shipped in a timely manner
- Specimen delayed in transport, by how long? _____ (days)
- Specimen lost in transport
- Other (explain)

Additional details if available:

Laboratory procedures

- Delay in running analysis
- Measurement error, Chemistry
- Measurement error, Instrument
- Misinterpretation of result
- Improper cutoff used
- Mishandling of specimen
- Wrong specimen assayed
- Abnormal value not recorded
- Misread identification number
- Clerical error
- Lack of notification of physician or follow-up coordinator
- Other (explain)

Additional details if available:

Follow-up

- No follow-up
- Second specimen requested but not received
- Follow-up coordinator unable to locate physician of record
- Follow-up coordinator unable to locate family of patient
- Follow-up coordinator delayed contacting physician or family of patient
- Other (explain)

Additional details if available:

Health provider practices

- Physician of record fails to notify patient or other physician of screening result
- Physician does not ensure that patient is retested in timely manner
- Physician does not order diagnostic tests
- Diagnostic results not communicated by laboratory to physician
- Diagnostic results not communicated by physician to patient
- After diagnosis, provider does not prescribe treatment
- Parent refuses retesting or treatment
- Other (explain)

Additional details if available:

Biologic variants

- Sample collected too early
- Late onset variant not detectable in first few days
- Form of disorder not detectable through assay used by screening laboratory
- Other (explain)

Additional details if available:

Other cause not identified in preceding list

Please explain:

Additional details if available:

5. Outcomes

Health outcome

- Death
- Mental retardation
- Developmental delay
- Other neurological symptoms (explain)
- Other (explain)

Additional details if available:

Legal outcome

Litigation

Yes No

Additional details if available:

6. Analysis

What steps could be taken to prevent this situation from recurring?
Please explain:

Has the state changed the newborn screening regulations, policies or protocols as a result of this case? Yes No
If yes, specify the changes:

7. Comments: