

7.3 Case Resolution

POLICY/GENERAL INFORMATION: Resolution of positives should occur as rapidly as possible to assure early diagnosis and treatment in the event that a child has the disease in question. While diagnoses of screened-for conditions can be made/confirmed only by physicians, ASC staff is responsible for assisting physicians in order to expedite the follow-up process (e.g., facilitating referrals to specialists, facilitating diagnostic testing, making referrals to CCS) and tracking/monitoring the status of the case to resolution. In the absence of outcomes documented in SIS by Special Care Centers, written documentation of the diagnosis and treatment shall be obtained and filed in ASC records.

Confirmatory Test Results

CHRCO, Stanford Laboratory, and the NAPS labs enter confirmatory test results for hemoglobin, biotinidase deficiency, and PKU, respectively. The Cystic Fibrosis Centers enter confirmatory results (sweat chloride tests). ASCs are to enter confirmatory test results for CAH, PCH, Bioppterin Deficiency, and Galactosemia in the “Confirmatory Test Results” screen. They do not have to enter confirmatory test results for the other metabolic MS/MS-positive cases.

- Hard copies of endocrine and non-MS/MS metabolic confirmatory results (or other documentation with lab results included) must be obtained. Cases may be closed in SIS prior to obtaining hard copies once a diagnosis is confirmed or ruled out, but the lab results must be entered when received. These confirmatory lab results should be entered into SIS by the ASC only after receipt of a hard copy of the results.
- In the event that a non-Quest lab is used for confirmatory testing for a metabolic disorder, a hard copy of lab results on which decision was based will be provided by the metabolic center directly to the MS/MS reviewer at the Genetic Disease Screening Program.
- Hemoglobin confirmatory testing should only be done at the state hemoglobin reference laboratory. If done at a non-contract lab, the ASC staff should contact GDSP Hemoglobin Coordinator and forward a hard copy of the results to him/her for review and entry into SIS.

Case Resolution/Closure

- Positives, and transfused cases that are being followed up, should not be closed until all the confirmatory test results, including DNA, have been entered into SIS by the respective confirmatory lab(s). If the ASC closes the case before this occurs, the lab can no longer access the case in SIS.
- For cases resolved as negative in the MSR or CFSR, or for those cases with negative confirmatory results that are entered in SIS by the state-contracted reference labs, written verification from the doctor is not required.
- Cases should be resolved as “Lost to Follow-up” when the whereabouts of the family are unknown and there is no way of communicating with them. This assumes that reasonable efforts to locate the family have been made and documented in SIS case notes. Before a case is resolved as such, it must be reviewed with the Project Director.

7.3 Case Resolution

- Cases should be resolved as “No Response” when ASC staff know the whereabouts of the family or have a method of communicating with them (e.g., via a relative or friend) but the family does not respond to calls, e-mails, letters, etc. This assumes that reasonable efforts have been made to communicate with the family and have been documented in SIS case notes. However, when this occurs, a referral to Child Protective Services must be considered (See Section 3.24). Prior to closing the case, the coordinator/program specialist must review the case with the Project Director. The case must be reviewed with the contract liaison before a referral to CPS is made.
- Cases should be resolved as “Refused” when parents inform ASC staff either verbally or in writing that they refuse to consent to further follow-up. However, when this occurs, a referral to Child Protective Services must be considered (See Section 3.24). Prior to closing the case as such, the issue must be reviewed with the Project Director. The case must be reviewed with the contract liaison before a referral to CPS is made.

Metabolic Cases Not Resolved within 6 Months; CF Cases Not Resolved within 9 Months

- For initial positive cases that metabolic centers have not resolved within 6 months of a baby’s life and the Cystic Fibrosis (CF) Centers have not resolved within 9 months of life, the ASC staff shall, after a discussion with the Project Director, reassign the cases to designated GDSP staff, who will continue to track the case until a resolution is reached and will enter the diagnosis/resolution in the SIS-Case Resolution screen.

Resolved Cases That Need to Be Reopened

- If a resolved case needs to be reopened for any reason (e.g., if the SCC retracts a diagnosis and changes the case status to “Pending,” requiring further ASC/GDSP follow-up; if a diagnosis needs to be changed), Contract Liaison needs to be notified so that change can be made in case registry. Tracking event 142 should be used to reopen case in SIS.

How to Resolve a Positive When a Baby Dies Before Confirmatory Testing Has Been Done/Completed

- If the PMD or specialist makes a diagnosis based on other evidence (e.g., symptoms), it can be resolved as Disease
- If the PMD or specialist hasn’t made a diagnosis but there is evidence to suggest the baby had the disorder, contact GDSP Nurse Consultant for direction.
- If there is insufficient information/evidence suggesting the disorder, resolve case as “Newborn Expired.”
- See Section 3.15.1 for specific information/instructions on expired babies with positive CF screens
- Update Client Profile with date of death and cause, if available.

How to Resolve a Positive When the Family Has Moved Out of State (or out of country) Before a Diagnosis Was Established or Ruled Out

- ASCs/GDSP Monitors are to make every effort to establish contact with the baby’s new PMD/Specialist to obtain follow-up and case resolution information. The case is not to be resolved as “Lost to Follow-up” unless the family’s new whereabouts are unknown.
- If ASC staff encounters difficulty in obtaining needed information, they should contact Contract Liaison, who will ask for assistance from that state’s NBS program staff.

7.3 Case Resolution

ATTACHMENTS/FORMS AND LETTERS TO BE USED:

3.20 A - Reporting Form for Confirmation or Rule Out of Screened-for Disorder

(To be completed by the ASC with as much information as possible prior to sending to physician.)

3.20 B - Cover Letter to Physician Reporting Diagnosis

3.20 C - Letter to Physician (Specialist) Informing Him/Her of Case Transfer

PROTOCOL:

METABOLIC POSITIVES

1. DISORDER RULED OUT

Resp. Person	Action
ASC Newborn Screening Coordinator or Program Specialist	<p>After <u>initial</u> MSR is entered into SIS by the metabolic center for either a phone consult or visit <u>and</u> after confirmation in SIS that a disorder is ruled out,</p> <ul style="list-style-type: none"> Resolves case as negative on Case Resolution screen in SIS. <p><u>*Exception:</u> Those Initial positives not requiring referrals to metabolic centers after the first screen (PKU, borderline positives for biotinidase deficiency, or galactosemia) do not require MSRs in order for the cases to be resolved as negative.</p>

METABOLIC POSITIVES

2. DISORDER DIAGNOSED BY CONFIRMATORY TESTING

Resp. Person	Action
ASC NBS Coordinator or Program Specialist	<p>After <u>initial</u> MSR is entered into SIS by the metabolic center for a phone consult or visit, diagnosis is confirmed, baby is seen by the metabolic specialist, <u>and</u> treatment plan is initiated, as indicated in the MSR:</p> <ul style="list-style-type: none"> Resolves with diagnosis, treatment start date, etc., on Case Resolution screen in SIS. If information is complete in MSR, states "See MSR for DX, RX info" in case notes. If information is not complete in SIS but has been provided to ASC via other means by metabolic center staff, enters information in case notes.

7.3 Case Resolution

METABOLIC POSITIVES

3. DISORDER NOT DEFINITELY RULED OUT OR DIAGNOSED AFTER CONFIRMATORY TESTING (pending additional work-up and testing by specialist)

Resp. Person	Action
ASC Newborn Screening Coordinator or Program Specialist	<ul style="list-style-type: none"> Monitors case in SIS (MSR) to confirm appointment with metabolic center. Contacts metabolic center/primary care physician(s) regularly (at least monthly unless informed via SIS or MD that baby will not be seen for over a month) to track status of case and documents findings in SIS. <p>If the case is not resolved on the MSR within 6 months of a baby's life,</p> <ul style="list-style-type: none"> Discusses case with Project Director Contacts specialist to inquire if a diagnosis had been made and inform him/her that ASC staff will no longer be tracking the case, but that state staff will be contacting him/her periodically to obtain required information about outcome of case Sends letter (3.20 C) to specialist stating same Enters a case note regarding current status of case Transfers case to designated GDSP staff (CCC101) via "Re-assign CCC" screen
GDSP Program Development and Evaluation Branch (PDEB) staff and Newborn Screening Branch (NBSB) staff	<ul style="list-style-type: none"> Monitors extended case list.
Designated PDEB MS/MS Monitor	<ul style="list-style-type: none"> Monitors MSR and contacts metabolic center periodically to obtain case resolution. When the specialist has resolved case as "Disorder" or "No Disorder" (as indicated in MSR) resolves case in SIS (Case Resolution screen).
NBSB staff	<ul style="list-style-type: none"> Provides technical assistance to PDEB monitor and/or metabolic center as needed.

7.3 Case Resolution

ENDOCRINE POSITIVES

1. DISORDER RULED OUT

Resp. Person	Action
ASC Newborn Screening Coordinator or Program Specialist	<p>After confirmation by physician that disorder is ruled out,</p> <ul style="list-style-type: none"> • Documents physician’s decision in SIS case notes. Includes name of physician. • Requests from physician: <ol style="list-style-type: none"> 1) Signed letter/other documentation (e.g., note written on lab report) stating decision, OR Completion of <i>Reporting Form for Confirmation or Rule-Out of a Screened-for Disorder</i>, OR Copy of either a history and physical, in-patient discharge summary, or medical record progress note with supporting documentation. 2) Hard copy of lab results on which decision was based unless lab results, reference ranges, and interpretation/diagnosis are included in above documentation. • Files documentation in baby’s record, enters confirmatory lab results into SIS, and closes on Case Resolution Screen.

ENDOCRINE POSITIVES

2. DISORDER DIAGNOSED

Resp. Person	Action
ASC Newborn Screening Coordinator Or Program Specialist	<p>After diagnosis made by physician <u>and</u> either:</p> <p>1) First appointment kept at CCS-approved special care center (SCC), if to be followed at a SCC OR 2) Patient has been seen and treatment/monitoring plan is initiated by PMD or specialist if not being followed at CCS SCC.</p> <ul style="list-style-type: none"> • Documents date of diagnosis, treatment information in SIS case notes. Includes name of physician. • Requests from physician: <ol style="list-style-type: none"> 1) Letter stating diagnosis, treatment, and date of initiation of treatment OR Completion of <i>Reporting Form for Confirmation or Rule Out of Genetic Disorder (3.20 A)</i>. OR Copy of a history and physical, in-patient discharge summary, or medical record progress note with needed documentation, e.g. diagnosis and treatment. 2) Hard copy of lab results on which decision was based unless lab results, reference range, and interpretation/diagnosis are included in above documentation. • Files documentation in baby’s record, enters confirmatory test results on Confirmatory Test Results screen, and completes Case Resolution screen.

7.3 Case Resolution

HEMOGLOBIN POSITIVES

1. DISORDER RULED OUT BY CONFIRMATORY TESTING

Resp. Person	Action
ASC Newborn Screening Coordinator or Program Specialist	After confirmation by confirmatory lab test that disorder is ruled out, <ul style="list-style-type: none"> • Resolves case as negative in SIS. Requests hard copy of lab results on which decision was based if done at non-contract lab. Contacts GDSP Hemoglobin Coordinator and forwards a copy of the results to him/her for review and entry into SIS. • Files documentation (not in SIS).

HEMOGLOBIN POSITIVES

2. DISORDER DIAGNOSED BY CONFIRMATORY TESTING

Resp. Person	Action
ASC Newborn Screening Coordinator Or Program Specialist	After diagnosis made by physician and either: 1) First appointment kept at CCS-approved special care center (SCC) , if to be followed at a SCC OR 2) Treatment/monitoring plan is initiated by PMD or specialist if not being followed at SCC. <ul style="list-style-type: none"> • Documents diagnosis and information above in SIS case notes. Includes name of physician • Requests from physician: <ol style="list-style-type: none"> 1) Letter stating diagnosis, treatment, and date of initiation of treatment. <p style="text-align: center;">OR</p> <p style="text-align: center;"><i>Completion of Reporting Form for Confirmation or Rule Out of Genetic Disorder (3.20 A)</i></p> <p style="text-align: center;">OR</p> <p style="text-align: center;">Copy of either history and physical, in-patient discharge summary, or medical record progress note with needed documentation, e.g. diagnosis.</p> 2) Hard copy of lab results if done at a non-contract lab. Contacts GDSP Hemoglobin Coordinator and forwards a copy of the results to him/her for review and entry into SIS. • Files documentation in baby's record. • Resolves with diagnosis on Case Resolution screen.

7.3 Case Resolution

CYSTIC FIBROSIS POSITIVES

1. DISORDER RULED OUT

Resp. Person	Action
ASC Coordinator or Program Specialist	<p>After initial CFSR is entered into SIS by the CF center for either a phone consult or visit, after confirmation in SIS that there has been confirmatory testing (sweat chloride test) and a disorder has been ruled out,</p> <ul style="list-style-type: none"> Resolves case as negative on Case Resolution screen in SIS.

CYSTIC FIBROSIS POSITIVES

2. DISORDER DIAGNOSED AS CF OR CRMS (CFTR-Related Metabolic Syndrome)

Responsible person	Action
ASC NBS Coordinator/Program Specialist	<p>After initial CFSR is entered into SIS by the CF center for either a phone consult or visit, confirmatory testing has occurred and the diagnosis is confirmed, baby is seen by a CF specialist (pulmonologist), and treatment plan is initiated, as indicated in the CFSR:</p> <ul style="list-style-type: none"> Completes Case Resolution screen. Once the CF center has resolved the case in the CFSR, screen will pre-fill with resolution, disease (CF or CRMS), date of diagnosis, name of specialist, date of treatment initiation). If information is not complete in SIS but has been provided to ASC via other means by CF Center staff, enters information in case notes.

CYSTIC FIBROSIS POSITIVES

3. DISORDER NOT DEFINITELY RULED OUT OR DIAGNOSED AFTER CONFIRMATORY TESTING (pending additional evaluation and testing by specialist)

Responsible person	Action
ASC NBS Coordinator/Program Specialist	<ul style="list-style-type: none"> Contacts CF center regularly (at least monthly unless informed via 'SIS or phone that baby will not be seen for over a month) to track status of case, and documents findings in SIS. <p>If after 9 months from accession date, the CF specialist has not arrived at a resolution:</p> <ul style="list-style-type: none"> Contacts the specialist to inquire if a diagnosis had been made and informs him/her that ASC staff will no longer be tracking case but that state staff will be contacting him/her periodically to obtain required case outcome information. Sends letter to specialist stating same. Transfers case to designated GDSP staff (CCC 102) via "Re-assign CCC" screen
<u>GDSP</u> Program Development and Evaluation Branch (PDEB) staff and NBS Branch (NBSB) staff	<ul style="list-style-type: none"> Monitors extended case list.
PDEB CF Monitor	<ul style="list-style-type: none"> Monitors CFSR and/or contacts CF Center periodically to obtain required information about outcome of case (i.e., CF diagnosed or ruled out) and documents findings in SIS. Once specialist has resolved case as "Disorder" or "No Disorder" in the CFSR, enters information in SIS on Case Resolution screen.
NBSB staff	<ul style="list-style-type: none"> Provides technical assistance to CF Monitor and/or CF center as needed.