### V.A. Research Evidence

**TABLE 3: Evidence Supporting Confirmatory Testing for Newborns with SCD**

<table>
<thead>
<tr>
<th>TYPE OF EVIDENCE</th>
<th>KEY FINDINGS</th>
<th>LEVEL OF EVIDENCE (USPSTF RANKING*)</th>
<th>CITATION(S)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Observational study</td>
<td>Vichinsky et al. followed two groups of children: 89 children diagnosed with SCD by a newborn screening program who were enrolled in a comprehensive care program and 64 children diagnosed with SCD after 3 months of age also participating in the comprehensive care program. The authors found that the mortality rate for patients diagnosed with SCD as a newborn was 1.8% compared with an 8% mortality rate among those diagnosed after 3 months of age. The lower mortality rate is attributed to early diagnosis and treatment of complications, which result in large part from extensive follow-up and patient and parent education.</td>
<td>II</td>
<td>Vichinsky E, Hurst D, Earles A, Kleman K, Lubin B. Newborn screening for sickle cell disease: effect on mortality. Pediatrics. 1988; 81(6):749-755.</td>
</tr>
</tbody>
</table>
| Clinical guidelines | **Recommendation to perform confirmatory testing:**

“For infants with positive screening tests, confirmatory testing of a second blood sample should be accomplished by 2 months of age so that parental education, prophylactic penicillin, and comprehensive care can be promptly implemented.” (NHLBI, p. 24)

**Recommendation to communicate results:**

“Results of the screen for SCD in the infant should be made available to the mother and father, as well as to the pediatrician.” (NHLBI, p. 164)

**Impact of confirmatory testing and communication of results on health outcomes:**

markedly reduces morbidity and mortality from SCD in infancy and early childhood.” (NHLBI p. 7)

| Clinical guidelines | In a 2000 statement from the Council of Regional Networks for Genetic Services, the council states, “the follow-up component [to a positive screening result] must ensure that the infant is located and brought into the diagnostic and management components of the system in time to ensure optimal outcome and to prevent irreversible damage” (Pass, pg. S11). In addition, the council states that, “Follow-up personnel should explain the test result [to the parent or guardian of the infant], recommend follow-up action according to the program protocol, describe the general principles of treatment, and answer any questions that arise so that follow-up action can be initiated in the appropriate time frame” (Pass, pg. S12). | III | Pass KA, Lane PA, Fernhoff PM, et al. US newborn screening system guidelines II: follow-up of children, diagnosis, management, and evaluation. Statement of the Council of Regional Networks for Genetic Services (CORN). J Pediatr. Oct 2000;137(4 Suppl):S1-46. |

**Note:** USPSTF criteria for assessing evidence at the individual study level are as follows: I) Properly powered and conducted randomized controlled trial (RCT); well-conducted systematic review or meta-analysis of homogeneous RCTs. II) Well-designed cohort or case-control analytic study. III: Opinions of respected authorities, based on clinical experience; descriptive studies or case reports; reports of expert committees.