

Sickle Cell Disease

Action required

Contact pediatric hematologist today.

Issues to discuss with hematologist

- Laboratory evaluation of infant
 - § Should testing be performed by primary care or hematology clinic
 - § Hemoglobin electrophoresis
 - § Sickledex is not appropriate
- Penicillin prophylaxis and other treatment
- Immunizations
- Prompt treatment of acute illnesses

Review with family

Family has **not** been notified of result by MDH.

After discussion with pediatric hematologist, contact family to coordinate clinic visit, lab work, and referral to hematology clinic. Generally, infants will be stable when family is contacted and at clinic visit.

Prompt follow-up is important. Family may need significant support to follow through with referral and treatment plan.

NICU issues

Newborn screens cannot be accurately interpreted after transfusion or in extremely premature babies, where adult hemoglobin cannot be detected.

Clinical summary

Sickle cell disease is an autosomal recessive disorder that results from the production of abnormal beta globin chains in red blood cells. These red blood cells sickle in states of low oxygen, dehydration, or stress/illness.

Infants are at significant risk for sepsis and pneumonia. Individuals with sickle cell disease face a life-long risk for hemolytic anemia, vaso-occlusive complications, infection, splenic sequestration, pain episodes, and stroke.

Children identified by newborn screening should receive prophylactic antibiotics, pain management, and should be monitored by both primary care and specialty providers.

Incidence: ~ 1/4,000; more prevalent in individuals of African descent (1:375)

Clinical expectations

If treated promptly, children with sickle cell disease can be expected to have near normal development.

Developmental delay, pain crises, and other symptoms can occur even with treatment.

Affected children should be monitored for pain, infection, and need for intervention.

Resources

GeneTests: www.genetests.org

OMIM: www.ncbi.nlm.nih.gov/sites/entrez?db=OMIM

ACT Sheets: www.acmg.net/resources/policies/ACT/condition-analyte-links.htm

MN Newborn Screening Program:
www.health.state.mn.us/newbornscreening