SICKLE CELL AND OTHER HEMOGLOBINOPATHIES

RATIONALE

Hemoglobinopathies are inherited conditions that cause disease by impacting the structure or amount of hemoglobin in the blood. If there is not enough hemoglobin in the body, or the hemoglobin is not formed correctly, oxygen cannot be carried through the body as efficiently.[1] If untreated, many of the inherited hemoglobin disorders result in death during the first few years of life.

There are many types of hemoglobin disorders and complications and they vary widely in terms of presentation and severity. Complications of hemoglobinopathies include anemia, pain crises, overwhelming infections, iron overload, splenic infarct, heart failure, kidney damage, and vascular compromise. While iron deficiency is the most common cause of acquired anemia, worldwide, hemoglobinopathies are the most common inherited causes of anemia. Yet, many individuals who are familiar with anemia caused by iron deficiency may not be aware that hemoglobinopathies can lead to anemia. For this reason, patients with hemoglobinopathies often go unrecognized or are misdiagnosed worldwide[4]. At this time no cure exists for these disorders, so the major approaches to the control and management of these diseases are population screening, genetic counseling, prenatal diagnosis, and management of symptoms.[2]

The rate of occurrence of hemoglobinopathies in California is more than 1 in 4,000 births. California has been screening for disorders of hemoglobin in newborns since 1990. This screening is part of the California's Department of Public Health Newborn Screening Program. This program screens for 80 conditions, including hemoglobin disorders. Early detection of hemoglobinopathies allows for early identification, treatment, and entry into comprehensive care centers as well as improved health outcomes for affected individuals (for further information please see the "Resources" section in this chapter to access a link to the California Newborn Screening Program).[3]

SCREENING REQUIREMENTS

- Verify that the California newborn screening test was completed and normal- this should detect most types of hemoglobinopathies, but may miss some of the rarer forms.
- If the provider is notified by the California Area Service Center (ASC) Newborn Screening Coordinator that a second blood sample called a "recall specimen" is necessary, the provider should collect it as requested.
- Request hemoglobin electrophoresis to test for hemoglobin abnormalities if a patient was not tested as a newborn and is anemic, or has failed to respond to iron replacement therapy for anemia.

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CONSIDERATIONS FOR REFERRAL TREATMENT AND/OR FOLLOW-UP

The California ASC Coordinator will assist the provider in referring a family diagnosed on newborn screening to a California Children's Services (CCS)-approved Metabolic, Endocrine or Sickle Cell Disease Center for specialized diagnosis and treatment[3].

- Children with hemoglobinopathies may have anemia despite adequate iron stores, they can develop iron overload, so it is important to rule out hemoglobinopathies in children who continue to have anemia despite iron supplementation.

Resources:

- National Center for Sickle Cell Disease and Comprehensive Thalassemia Center at UCSF Benioff Children's Hospital Oakland
- California Newborn Screening Program
- CCS approved Sickle Cell Disease Centers
- Anemia and Red Blood Cell Disorder Team at UCLA Children's Hospital

References:

[1] Kansas Department of Health and Environment. Hemoglobin Disorders (Hemoglobinopathies) Information for Parents. Kansas Department of Health and Environment. Published May 20, 2011. Accessed August 28, 2014.

[2] Weatherall D, Akinyanju O, Fucharoen S, Olivieri N. Inherited Disorders of Hemoglobin. In: Jamison DT, Breman JG, Measham AR, et al., 2nd edition. *Disease Control Priorities in Developing Countries*. Washington (DC): World Bank; 2006: Ch. 34.

[3] California Newborn Screening Program. How Common Are These Disorders in California. California Department of Public Health. Last Modified June 15, 2011. Accessed August 28.2014.

[4] Family Health Services Division Children with Special Health Needs Branch, Genetics Program. Thalassemias and Other Hemoglobinopathies Protocol Hawai'i. Hawai'i Department of Health. Published January 2008. Access August 28, 2014.

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