NERGG, Inc.
New England Regional Genetics Group

Public Health Genomics Task Force:
A working group of NERGG, Inc.

Genomics Alert: an Update for Chronic Disease and Public Health Staff

The New England Regional Genetics Group (NERGG) Public Health Genomics Task Force would like to encourage and support your efforts in educating the public about chronic disease conditions which may have a genetic component. Individuals who are aware of their family histories have an opportunity for early diagnosis and intervention, enabling them to institute healthy life styles and preventive measures which will enable them to be as healthy as possible. To that end, we summarize and share related information in this format with chronic disease program staff and other interested parties for their outreach and advocacy use. These Genomics Alerts will be disseminated periodically, please share them with others who may be interested.

We would appreciate your feedback on this Alert, as well as your suggestions for future topics. Please feel free to contact your state's Task Force representative with your feedback. To be added to the distribution list, contact Mary-Frances Garber/NERGG Executive Director at mfgnergg@verizon.net. *Thank you for your interest in public health genomics education.*

September is Sickle Cell Disease Month

What is Sickle Cell Disease (SCD)?

Sickle cell disease (sometimes known as sickle cell anemia) is a group of life-long blood disorders that affect hemoglobin, the molecule in red blood cells that delivers oxygen to cells throughout the body. SCD occurs from genetic changes which cause a portion of the hemoglobin molecules to be abnormal and the red blood cells to be "sickle"-shaped rather than round. When sickle-shaped, the cells can become stuck in blood vessels and prevent blood from flowing properly throughout the body. These blockages can cause episodes of pain, organ tissue damage, infections, or stroke.

Sickle cell disease is an inherited condition. In order to have sickle cell disease, a person must inherit a genetic mutation within the sickle cell gene from each of their parents. A person who receives an altered gene for SCD from one parent and a normal form of the hemoglobin gene from the other has a condition called "sickle cell trait." Most people with sickle cell trait do not exhibit symptoms of the disease.

The Public Health Burden of Sickle Cell Disease

SCD is the most common inherited blood disorder in the United States. The Centers for Disease Control and Prevention and the National Institutes of Health estimate that[1,2]:

- About 1,000 babies are born with SCD each year in the United States.
- The genetic blood disorder affects 70,000–100,000 Americans, the majority of whom are African American or Hispanic.
- Sickle cell disease occurs in approximately one out of every 500 African American births and one out of every 36,000 Hispanic American births, but can occur in people of many ethnic origins. For this reason, screening all newborns for the disorder is recommended.
- About 2.5 million people in the United States have sickle cell trait.

Who is affected by SCD?

SCD affects millions of people throughout the world and is particularly common among those whose ancestors came from sub-Saharan Africa; Spanish-speaking regions in the Western Hemisphere (South America, the Caribbean, and Central America); Saudi Arabia; India; and Mediterranean countries such as Turkey, Greece, and Italy.

SCD and Family History

Children are born with SCD only when they inherit the sickle cell form of the gene from both parents. It is important to know if you or a family member has sickle cell disease or sickle cell trait, particularly when planning a family. Early identification and treatment of SCD can help prevent and avoid complications.

If someone in your immediate family has SCD or sickle cell trait, or if your ancestors are from a part of the world where SCD is common, you could be a sickle cell carrier (i.e., have sickle cell trait) and your future offspring may be at increased risk for SCD, in which case you and your partner may want to seek genetic counseling and/or testing for your carrier status either prior to conception or during pregnancy.

Resources for Outreach and Prevention Efforts

- Sickle Cell Disease, CDC: http://www.cdc.gov/ncbddd/sicklecell/index.html
- What is Sickle Cell Anemia? Includes videos of "Our Story: Living With and Managing Sickle Cell Disease"; NIH, National Heart, Lung, and Blood Institute: http://www.nhlbi.nih.gov/health/health-topics/topics/sca/
- Sickle Cell Disease Association of America: http://www.sicklecelldisease.org/
- **Tools to integrate family history into your programming:**
    - My Family Health Portrait tool. US Surgeon General, at: https://familyhistory.hhs.gov/fhh-web/home.action
    - Family Health History Pocket Guide, Workbook and Poster. Produced by the Connecticut Department of Public Health Genomics Office, available at: http://www.ct.gov/dph/cwp/view.asp?a=3134&q=387816&dphNav_GID=1822&dphNav_GID=1822
    - Does It Run In the Family? On-line Customizable Toolkit (http://www.familyhealthhistory.org/): Allows users to create customized versions of two family health history booklets, "A Guide to Family Health History" and "A Guide for Understanding Genetics and Health." Booklets can tailored to include personal health stories, pictures, interview questions, disease information, etc.

References

1. Sickle Cell Disease, CDC. http://www.cdc.gov/ncbddd/sicklecell/. Accessed July, 2012.
2. National Heart, Lung, and Blood Institute, National Institute of Health. http://www.nhlbi.nih.gov/health/health-topics/topics/sca/atrisk.html. Accessed July, 2012.