Ambry Genetics Launches New Genetic Test for Hemoglobin Disorders

IRVINE, CA (February 11, 2005) - AMBRY GENETICS, a leading genetic testing company, announced it has launched a new test to detect any mutation within the coding region of the beta globin gene (HBB) and the surrounding intronic sequences. The Ambry Test: Beta Globin aids in the diagnosis of hemoglobinopathies and provides a solid basis for proper therapy.

Disorders involving hemoglobin are among the most common genetic disorders worldwide, with approximately 5 percent of the world's population being carriers for clinically important hemoglobin mutations. Genes involved are those that control the production of proteins known as globins, contained in hemoglobin. Adult hemoglobin is composed of 2 alpha and 2 beta chains. Hemoglobin contained within the red blood cells binds oxygen reversibly and controls the cell's capacity to transport oxygen to the tissues. Approximately 500 beta globin mutations have been discovered, many of which cause serious clinical effects.

Mutations in the DNA sequences controlling beta globin synthesis can:

produce structurally abnormal hemoglobins (e.g., sickle-cell anemia)

diminish production of the hemoglobin molecule (e.g., thalassemia)

impair the developmental modification from fetal to adult hemoglobin

Sickle Cell Disease

The term sickle cell disease encompasses a group of symptomatic disorders associated with mutations in the HBB gene. Sickle cell disease is inherited in an autosomal recessive manner and causes red blood cells to become deformed (sickle-shaped).

Approximately 80,000 Americans have sickle cell disease. HBB alleles associated with sickle cell disease are common in African, Mediterranean, Middle Eastern, and Indian ethnicities as well as in persons from the Caribbean and parts of Central and South America. In the US, 1 out of 10 African Americans have sickle cell trait and 1 out of 625 newborns are affected with the disease.

Thalassemia

Beta-thalassemias are caused by mutations in the beta globin gene that result in decreased or absent production of beta globin proteins. When a patient has had an abnormal hematological analysis, molecular genetic testing of the HBB gene can be performed by Ambry Genetics in order to identify the disease-causing mutation(s). Identification of specific beta globin mutations can confirm a differential diagnosis and improve care for the patient as well as for the patient's family.

Most people who inherit thalassemia are Asian, Filipino, Mediterranean, Middle Eastern, or of African descent. b-thalassemia mutations are population specific: each ethnic group has its own subset of common mutations. The Ambry Test: Beta Globin removes the bias and limitations of other tests by analyzing the entire gene and detecting all mutations that may be present.

Ambry Genetics' Beta Globin analysis is the most cost-effective, comprehensive primary test method for thalassemia and can also be used following inconclusive hemoglobin electrophoresis screening. Early and accurate diagnosis of b-thalassemia is currently the best tool for decreasing morbidity and mortality from this disease.

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The "New Products" page is designed to offer you news and information from businesses serving the genetics community. We welcome your submissions. All submissions are subject to review by the Editor. For more information, contact Al Lucchesi, National Accounts Manager, Lippincott Williams & Wilkins, 530 Walnut Street, Philadelphia, PA 19106; phone 215-521-8409; fax 215-521-8411; email alucches@lww.com.