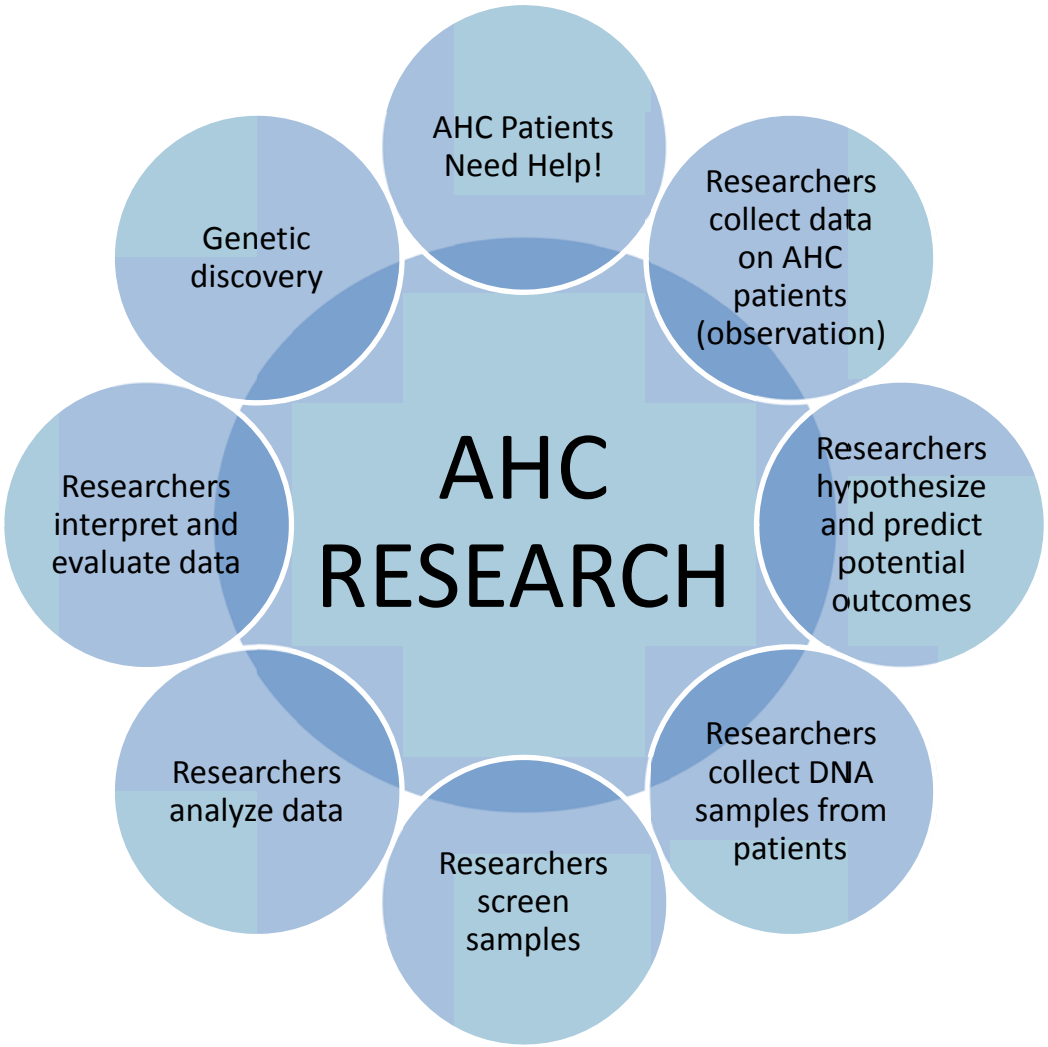
IT ALL BEGINS WITH THE AHC PATIENT
And The Scientific Process



AHC FAMILIES NEED HELP UNDERSTANDING GENETICS

Are you a little confused by all this talk about "The Gene Discovery?"

Do you wish you had a simple way of understanding why DNA and genes are so important to understanding AHC?

Do you know how to get tested for the gene mutation?

The Alternating Hemiplegia of Childhood Foundation is here to help.
We are pleased to provide you with this guide on understanding several basic points on AHC and genetic research.

We hope this guide will help you feel comfortable asking questions of your physicians and talking about the latest scientific discoveries around AHC.

UNDERSTANDING BASIC GENETIC DEFINITIONS

PROTEINS

Proteins are large, complex molecules that play many critical roles in the body. They do most of the work in cells and are required for the structure, function, and regulation of the body's tissues and organs.

CELLS

Cells are the basic building blocks of all living things. The human body is composed of trillions of cells. They provide structure for the body, take in nutrients from food, convert those nutrients into energy, and carry out specialized functions.

DNA

DNA, or deoxyribonucleic acid, is the hereditary material in humans and almost all other organisms. Nearly every cell in a person's body has the same DNA. Most DNA is located in the cell nucleus (where it is called nuclear DNA), but a small amount of DNA can also be found in the mitochondria.

GENES

A gene is the basic physical and functional unit of heredity. Genes, which are made up of DNA, act as instructions to make molecules called proteins. In humans, genes vary in size from a few hundred DNA bases to more than 2 million bases. The Human Genome Project has estimated that humans have between 20,000 and 25,000 genes.

CHROMOSOMES

In the nucleus of each cell, the DNA molecule is packaged into thread-like structures called chromosomes. Each chromosome is made up of DNA tightly coiled many times around proteins called histones that support its structure.

Source: http://ghr.nlm.nih.gov/handbook/

THE BASICS OF GENETIC TESTING AND THE A

What is Genetic Testing?

It is a type of medical test that identifies changes in chromosomes, genes, or proteins. The results can confirm the existance of a genetic disorder.

What methods of genetic testing can be used to help identify variations or mutations that lead to a genetic disorder?

Molecular Method

tests single genes or short lengths of DNA

Chromosomal Method

analyzes whole chromosomes or long lengths of DNA

Biochemical Method

studies the amount or activity level of proteins

How does A

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Text is from http://ghr.nlm.nih.gov/handbook/testing?show=all