What Causes Mitochondrial Disease?

Mitochondrial diseases aren’t contagious are caused by mutations, or changes in the DNA that make up a person’s genes. Genes are the cells’ blueprints for making proteins, which are used to build cells and structures of the body, like muscles and hair. Mitochondrial disorders occur when the cell’s mitochondria don’t work properly, which can result in various symptoms. In some cases, the disease is fatal in early childhood. In other cases, a person can live with a mitochondrial disorder with only minor symptoms. Some of these disorders are caused by mutations in the nuclear DNA (nDNA) of the individual, but some are caused by mutations in the mitochondrial DNA (mtDNA). Overall, mitochondrial disorders are complex because they involve multiple genes and multiple proteins. Changes in these proteins then result in physiological symptoms in the individual.

How Do Mitochondria Work?

Each mitochondrion is an energy factory that “imports” sugars and fats, breaks them down and “exports” energy in the form of ATP. This process is called cellular respiration.

The genes involved in mitochondrial disease make proteins that work inside the mitochondria. Within each mitochondrion, these proteins make up part of an assembly line that uses food molecules to manufacture the energy molecule, adenosine triphosphate (ATP). Proteins toward the end of the line are organized into five groups called complexes: I, II, III, IV and V. A mutation can cause a problem with any of these protein complexes. Any one of these complexes can be associated with a mitochondrial disorder, resulting from a mutation in the DNA that codes for that particular protein.

A deficiency in one or more of these complexes is the typical cause of a mitochondrial disease. In fact, mitochondrial diseases are sometimes named for a specific deficiency, such as complex II deficiency. You can find a list at the United Mitochondrial Disease Foundation (http://www.umdf.org/types/) Because there are so many possible mutations, and proteins that can be affected, there are many different types of mitochondrial disorders and outcomes. Some changes may only cause minor disfunction, and the cells can produce enough ATP to power body functions. In other cases, the broken protein results in the cells not producing enough ATP to satisfy the needs of the body.

When a cell is filled with defective mitochondria, not only does it become deprived of ATP, it also can accumulate unused glucose and oxygen. Cells of the brain and muscles are very sensitive to energy needs and are seriously affected by lack of ATP. Children can suffer from encephalomyopathy and developmental delays. Muscles may become weak (hypotonia) or they may lose the ability to move at all (ataxia). Parents may notice a variety of problems with their infants before getting an official diagnosis. Unfortunately, there is no cure for mitochondrial disorders.
1. Vocabulary: Write a short definition or description for each of the following terms.

mutation

cellular respiration

ATP

encephalomyopathy

nDNA and mtDNA

protein complex

2. What is the CLAIM (or main idea)?

3. What is the EVIDENCE presented for this claim?