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Short Communication

Comprehensive Mitochondrial Amyotrophic Lateral Sclerosis Disorder: Genetics to Genes

Samuel Smith*

Department of Molecular Medicine, Murdoch University, Murdoch, Australia

DESCRIPTION

Mitochondrial disorders are a type of metabolic ailment that affects mitochondria. Mitochondria are tiny structures found in almost all of your cells that make energy. They generate it by coupling oxygen with food-derived fuel molecules (sugars and fats). When the mitochondria are damaged, the cells do not have adequate energy. Mitochondrial diseases are long-term (chronic), hereditary and sometimes inherited conditions in which mitochondria fail to provide enough energy for the body to function normally. Mitochondrial disorders can affect cells in the brain, nerves, muscles, kidneys, heart, liver, eyes, ears, and pancreas, among other places. Mitochondrial dysfunction happens whenever the mitochondria are unable to function properly due to some other particular disease. Secondary mitochondrial malfunction can be caused by a variety of disorders, including Alzheimer's disease, muscular dystrophy, Amyotrophic Lateral Sclerosis (ALS), Diabetes, and cancer [1].

Mitochondrial dysfunction

Amyotrophic lateral sclerosis, also known as ALS, is a degenerative nervous system disorder that impacts both upper and lower motor neurons in the brain. Lou Gehrig's sickness is named after the baseball player who was diagnosed with it. ALS symptoms include muscle twitching and weakness in a limb, as well as slurred speech. ALS eventually affects the muscles that allow you to move, speak, eat, and breathe. This deadly condition has no known cure. Inside cells, mitochondria are small energy "factories." They resemble microscopic cells and contain their own DNA. Mitochondrial dysfunction may play a role in the development and progression of ALS. Mitochondria are organelles that play a key role in eukaryotic cell metabolism and survival, such as ATP generation, phospholipid biosynthesis, calcium homeostasis, calcium signaling, and apoptosis [2,3]. About 1500 proteins are involved in various mitochondrial functions; the majorities are encoded by nuclear genes, although tiny percentages are encoded by Mitochondrial DNA (mtDNA). Furthermore, one of the consequences of mitochondrial dysfunction is decreased energy production, which has a significant impact on ALS neurons because neuronal activity and synaptic function are dependent on the energy supply provided by mitochondrial Oxidative Phosphorylation (OXPHOS) complexes [4].

Genetics behind the ALS

The word "familial" ALS refers to the disease affecting multiple members of a family. Once someone has developed an ALS-causing mutation, his or her children can inherit it, making their disease "familial." ALS-causing mutations are predicted to exist in at least 25 distinct loci. The plural version of locus refers to the location on a chromosome of a gene or other important sequence. Although most cases of familial ALS are autosomal dominant, recessive and X-linked variants have been identified. The term "autosomal" refers to a mutation that arises on a chromosome other than X or Y. Dominant indicates that a disease can be caused by only one copy of a gene (usually, a person has two copies of each gene, one inherited from the father and one acquired from the mother). A person, who gets a defective gene from a parent, as well as the parent, will develop the disease. Two copies of a faulty gene are required to manifest the disease in a recessive inheritance pattern. Each parent passes on one copy of the gene, but neither parent has the condition. The genetic flaw (or mutation) in an X-linked form of ALS is found on the X chromosome [5].

CONCLUSION

The most common mutations are in Superoxide Dismutase-1 (SOD1), Chromosome 9 Open Reading Frame 72 (C9ORF72), TAR DNA/RNA-binding protein of 43 kDa (TARDBP), and RNA-Binding Protein Fused in Sarcoma (FUS), though other genes with very low mutation frequencies or even private mutations have been documented. The most prevalent genetic causes of ALS are mutations in the superoxide and frame 72 genes. However, as previously stated, additional genes are linked to the condition. Superoxide dismutase 1 gene mutation is linked

Correspondence to: Samuel Smith, Department of Molecular Medicine, Murdoch University, Murdoch, Australia, E-mail: Smith_s007@hotmail.com

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linked to ALS1. The superoxide gene codes for the superoxide dismutase enzyme, which is found in abundance in cells throughout the body. To break down harmful, charged oxygen molecules termed superoxide radicals; this enzyme binds to copper and zinc molecules. The mutations in the superoxide gene were discovered to be the cause of familial ALS. Since then, scientists have found many more faulty mitochondrial genes that can cause familial ALS. The C9 or F72 repeat expansion is transcribed both in the sense and antisense pathways, resulting in RNA foci containing repeats in patient tissues. The development of RNA foci aids the recruitment of RNA-binding proteins, induces mislocalization, and disrupts their normal functioning.

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