Fatal Familial Insomnia Essay, Research Paper

Fatal Familial Insomnia

Fatal familial insomnia is a genetic disorder. It manifests itself by many symptoms due to the degeneration of a certain part of the brain, the thalamus. The disease also results in the formation of amyloid plaques. This is the build up of a waxy substance made of proteins associated with polysaccharides. The disease is a result of a mutation of a normal protein that is associated with brain tissue. This is the prion protein. In the case of fatal familial insomnia, the mutation occurs 178 amino acids into the normal protein. Were an asparagine should be, an aspartic acid is instead. This disease is an autosomal dominant, which means that both sexes are affected and there are no carriers. If an individual inherits the mutant gene, that individual will at some point suffer the disease.

In the case of fatal familial insomnia, the affected area of the brain is the area responsible for sleep, the thalamus. The thalamus is the center which communications from the brain to the body and the body to the brain pass through for proper directions to where a signal should be received. When sleep takes place, it is thought that the thalamus becomes less efficient at this signal transfer function allowing for the vegetative state of sleep to come over an individual. Consequently, the symptoms of fatal familial insomnia are directly related to the malfunction of the responsibilities of the thalamus, namely sleep. Sleep, blood pressure, heart rate, body core temperature and hormone flow are all affected by the interruption of the body’s circadian rhythms which is a direct result of the degeneration of the thalamus in this disease. Other symptoms of this disease include the inability to produce tears or feel pain as well as poor reflexes and dementia. The lack of sleep leads to other problems such as hallucinations and coma. This is a clear demonstration of a pleiotropic disease, a disease with many phenotypic expressions. That is, this disease is the result of one mutant gene yielding one mutant protein, yet causes many physical abnormalities such as skin blotches, lack of tears, etc.

In the case study of an Italian family where of 288 relatives over 6 generations, 29 are affected by the disorder. The average age of onset of the disease is 49, but this may vary with the individual as with one female who was 61 years of age. Her disease lasted 18 months and followed the following pattern of the disease.

There are four stages of the disease before an individual’s life ends. The first stage is progressive insomnia, the trade mark of fatal familial insomnia. The first stage develops over approximately four months and includes a collection of psychiatric problems such as panic attacks and bizarre phobias. The second stage includes hallucinations, panic, agitation and sweating and lasts about five months. The third stage lasts about three months and is total insomnia with weight loss. The individual at this point looks much older and may experience incontinence. The fourth stage is around six months long and is recognized as dementia, total insomnia and sudden death after becoming mute.

This disease does not show until or past child bearing years when potentially affected individuals may have already had children that may also be potentially affected. Because of this fact, modern biotechnology must be employed for early diagnosis. Techniques such as DNA sequencing or molecular hybridization with a probe which seeks to detect the defective gene may be used for early diagnosis.

As for the treatment of this disease, hope may be found in the advancement of something called gene therapy. This treatment involves the insertion of the correct gene into an affected individual altering his/her gene expression making it what it should be for the expression of the correct protein. In order for this to happen, early diagnosis of an individual must be accomplished, possibly by the mentioned biotechniques above. This is so that the defective gene may be repaired before the onset of the disease. In order for this to be possible, the corrective gene must be isolated. Furthermore, the corrective gene must be good for transfer as well as a the proper vector to effectively execute the transfer. Because there is no cure for this illness, gene therapy may be the only answer if it is one day successful.

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Researchers discover new form of insomnia

Researchers have discovered a brain-wasting disease that begins with severe insomnia and ends in hallucinations and death.

The disease, sporadic fatal insomnia, is caused by the same type of deformed proteins, known as prions, that cause mad cow disease and its human variant, Creutzfeldt-Jakob disease, according to two studies.

There is no known treatment or cure. It is not clear what causes the disease, but scientists know it is not inherited. And unlike one type of Creutzfeldt-Jakob disease, it is not believed to come from infected meat.

Instead, scientists suspect it is caused by a spontaneous mutation in a single brain or nerve cell.

So far, they have identified only six cases of the disease, but there could be others that were misdiagnosed as other mind-destroying illnesses such as Alzheimer’s disease.

Prions are proteins with Jekyll-and-Hyde personalities that cluster in the brain. When their molecules are folded into the correct shape, prions are benign, though no one knows their function.

Folded the wrong way, prions induce other proteins to mimic them. The misfolded prions then accumulate in parts of the brain, causing the tissue to break down and become full of holes, like a sponge.

Different prion diseases attack different parts of the brain, causing characteristic types of dementia and death.

Some are infectious, like mad cow disease in cattle and scrapie in sheep and goats. In humans, prion diseases are either inherited, caught from eating contaminated meat, or spontaneous.

Researchers have previously identified an inherited prion disorder whose main symptom is sleeplessness, called fatal familial insomnia.

In a case outlined in Thursday’s New England Journal of Medicine, a 44-year-old patient had all the symptoms of the inherited disease, including prions with the same ‘’signature,” but did not have the inherited mutation. The case was reported by Dr. James Mastrianni, a neurologist at the University of Chicago.

The spontaneous, or sporadic, versions of the prion diseases are rare, but more common than their inherited counterparts.

Prions are still controversial. Some researchers believe it is impossible for proteins to replicate and cause disease, since they do not contain genetic material. These researchers believe the diseases are instead caused by as-yet-undiscovered viruses.

However, the new studies add to evidence the infection is caused by the abnormal shape of the prions, not another agent, Drs. Pierluigi Gambetti and Piero Parchi of Case Western Reserve University said in an accompanying editorial.

Gambetti and Parchi have described five similar cases of non-inherited fatal insomnia in a study scheduled to appear in the journal Neurology.