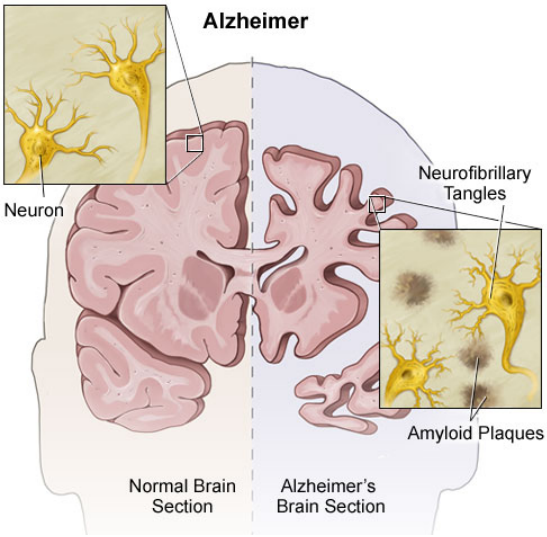
Mutations and the Brain



http://www.personal.psu.edu/users/j/n/jnb5091/Images/CFTR%20

Text Mark Up Directions:

1. Preview and **number** the paragraphs.
2. Read once and **circle the vocabulary** terms; then look them up and **define** or write a synonym in the margin next to the word.
3. Re-read a second time and **highlight** the main points (per paragraph) and **annotate** (paraphrase) in the margin.

Lois noticed her always jovial co-worker Christina, was beginning to be irritable and argumentative. She was often forgetting conversations they'd had and was having difficulty remembering client information at work-something she had never done. Christina was diagnosed with early onset Alzheimer's at age 52 just before she was ready to retire. Within three years she began having difficulty remembering even her husband's name and saying the simplest sentences.

Her brain was shrinking due to malfunctioning connective proteins that were shriveling into clumpy masses instead of making the linking tentacles that help to provide memory connections. These clumpy plaques were creating voids in her brain where neuron cells died after becoming disconnected and starved for nutrients leaving Christina with memory loss. The process continued to worsen until it reached her brain stem and her brain forgot how to control swallowing and breathing. Christina passed away in three short years after her first diagnosis with Alzheimer's.

All this damage in her brain was due to a single gene mutation on one of three possible chromosomes 1, 14 or 21; all of which contained genes controlling brain cell connectivity. A single DNA nucleotide change could result in a faulty protein by changing the genetic recipe and producing an incorrect amino acid. After translation, the order of the amino acids in the protein chain will cause the chain to fold up into precise shapes which suit their function. When the amino acids were changed the proper folding didn't take place and as a consequence her connective proteins failed to work.

A child whose parent has a faulty gene will have a 50/50 chance of inheriting it but only an increased *likelihood* of developing the disease. Weather or not that particular gene is expressed or "switched" on/off can be affected by environmental factors at any time in life. These factors include stress hormones, lack of exercise, molecules in a poor diet, environmental chemicals, or smoking, to which an individual may be exposed, even in the womb. These activities can alter a cell's DNA in ways that affect the activity of genes and make people more or less susceptible to developing a disease. The emerging new study of how lifestyle environmental factors affect DNA and disease is called epigenetics.

Follow Up Questions:

1. What were some of the outward symptoms or signs that Christina was sick?

2. What happened to the brain cells in Christina's brain?

3. How did a nucleotide change affect her connective proteins?

4. What are some epigenetic factors that may alter DNA during a person's lifetime?