CALIFORNIA STATE SCIENCE FAIR
2004 PROJECT SUMMARY

Name(s)
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Project Number
J0411

Project Title
Swimming Backstroke in the Gene Pool

Abstract

Objectives/Goals
Scientists have found a gene that may play a part in whether an athlete is a sprinter or excels at endurance events. This gene has been named ACTN3 and it makes a protein called actinin-3 that helps muscle cells re-power quickly. A change (mutation of C to T) in ACTN3 can cause the cell to stop building the actinin-3 protein, leaving it useless to the muscle cell. Without actinin-3, however, the athlete may be better at long distance events. Genes that are important to our survival as a species are also seen in animals related to us through evolution. Scientists have found the ACTN3 gene in other animals, but not the change (mutation of C to T) that makes its protein absent from the muscle cell. The goal of this science project is to see if the C to T change in ACTN3 is found only in humans or if it is found in other primates as well.

Methods/Materials
Polymerase Chain Reaction (PCR), which makes many copies of the DNA of interest, coupled with DNA sequencing, was used to tell if the C to T change was found in the ACTN3 gene. General Materials: micropipets, pipettors, microcentrifuge tubes, test tube racks, centrifuge, ice bucket/ice. PCR: DNA, primers, DNA polymerase, deoxynucleotides, tris buffer, water, PE 9700 PCR machine. Gel Electrophoresis: agarose, tracking dye, gel apparatus, power supply. Gel Sraining: pyrex dishes, ethidium bromide stain, water. DNA Sequencing: PCR product, deoxynucleotides, terminator nucleotides, DNA polymerase, water, ABI 3100 DNA Sequencer

Results
Out of 5 human samples, three have both C and T at ACTN3, and two have only T. Out of 25 primate samples, 15 have only C, seven have both C and T, and three have only T.

Conclusions/Discussion
The mutation (C to T) in the ACTN3 gene is found in primates as well as humans. I conclude that this mutation is not specific to humans, and likely originated millions of years ago. Usually, when a mutation causes the cell to stop building a protein the result is disease or even death. This protein stopping mutation in ACTN3, however, seems to be good, since many primates (including humans) have kept it.

Summary Statement
I determined that a mutation that may play a part in whether an athlete excels at endurance events originated millions of years ago.

Help Received
This research was conducted in my mother and father's laboratory at the University of California, Irvine Medical School. Simin Hakim helped me run my PCR and DNA sequencing reactions.