



**CALIFORNIA STATE SCIENCE FAIR  
2004 PROJECT SUMMARY**

<b>Name(s)</b> <b>Rebecca L. Kaspar</b>	<b>Project Number</b> <b>J0407</b>
<b>Project Title</b> <b>GENEalogy: Tracing a Mutant Gene through My Family Line</b>	
<p style="text-align: center;"><b>Abstract</b></p> <p><b>Objectives/Goals</b> The purpose of this study was to find out how many of my relatives have a prothrombin gene mutation that my father carries. Do my relatives and I also carry the mutant gene, and if so, is the mutation passed on from parent to child? I hypothesized that 50 percent of my family members would have the prothrombin mutant gene.</p> <p><b>Methods/Materials</b> Materials: 50 ml and 1.5 ml tubes, Cheek cells, PBS, dNTPs for Taq polymerase, Taq polymerase, Thermocycler, Agarose gel, Buffer with salt, DNA standards, HindIII enzyme, Centrifuge, DNA size markers, Ethidium bromide, Electrophoresis apparatus, Ultraviolet light, DNA oligonucleotides, Loading dyes. Methods: Swish mouth with PBS. Isolate the DNA. Make the agarose gel. Set up Polymerase Chain Reaction (PCR). Cut with HindIII. Separate the DNA fragments on the agarose gel until the dye is almost to the bottom. The gel showed one or two bands, depending on whether the piece of DNA had been cut once (wildtype) or twice (mutant); this showed that the person had the mutant gene or if both genes were wildtype.</p> <p><b>Results</b> The results showed that my grandfather, 4 of 6 aunts and uncles, and 5 of 13 cousins all had the prothrombin gene mutation. My fathers mutation was not a spontaneous mutation and was a dominant gene.</p> <p><b>Conclusions/Discussion</b> In conclusion, I discovered that 48 percent of my family carry the prothrombin mutation, which proved my hypothesis.</p>	
<b>Summary Statement</b> I traced a prothrombin gene mutation through my family line.	
<b>Help Received</b> Roger Kaspar drove me to the lab many times and taught me the correct use of the equipment. Brian Johnston provided the equipment. BioRad and SomaGenics donated some of the materials.	