



**CALIFORNIA STATE SCIENCE FAIR
2010 PROJECT SUMMARY**

Name(s) Kevin C. Yang	Project Number S0422
--	---------------------------------------

Project Title
Mitochondrial DNA, Our Second Genome: Unique Haplotypes of Mitochondrial DNA Control Region in a Han Chinese Family

Abstract

Objectives/Goals
Mitochondrial DNA (mtDNA)'s control region accumulates mutations at approximately 10 to 20 times the rate of nuclear DNA. However, mtDNA from Han Chinese in the Shanghai-Zhejiang region has been poorly sampled and understood in its variation, with only one paper published (Nishimaki et al., 1999). The purpose of this study is to examine how the mtDNA sequences differ among my family members and whether my family members have unique haplotypes of mtDNA control region compared to the people of my family's origin.

Methods/Materials
mtDNA was purified by boiling mouth cheek cells in the presence of 10% Chelex and centrifugation. PCR amplified 1070 bp fragment was digested using the enzyme MseI and analyzed by agarose gel electrophoresis. PCR amplified 440 bp fragment was sent to DNA Learning Center, Cold Spring Harbor Laboratory for sequencing. This sequence was then analyzed and compared against Revised Cambridge Reference Sequence (rCRS) and against each other using CLUSTAL W.

Results
5 sequence polymorphisms were observed in the mtDNA control region of the father when compared to that of rCRS. They were at nucleotide position: 16184, 16223, 16293, 16298, and 16319. 4 sequence polymorphisms were observed in the mtDNA control region of the mother and children when compared to rCRS. They were at nucleotide position: 16129 16182 16183 and 16189. Father does not have the Mse I restriction site in his mtDNA control region at nucleotide position 16297, consistent with the sequencing finding that there is a T to C polymorphism at 16298 which interrupts the TTAA Mse I cutting site.

Conclusions/Discussion
Father's mtDNA control region sequence is different from that of the mother and children. Mother's DNA sequence is the same as those of the children; consistent with the theory that mtDNA is passed maternally. Although new mutations may occur in the children, this was not observed in the mtDNA sequences of the children. Neither haplotype of our family was described in Nishimaki's paper. Therefore, this study found unique haplotypes of mtDNA control region in the family which was never described in the Shanghai-Zhejiang region before.

Summary Statement
Mitochondrial DNA's control region from members of a family was analyzed and unique haplotypes were found that were never described previously in the family's homeland.

Help Received
I gratefully acknowledge my family members for donating the samples and ordering all the reagents and equipments; DNA Learning Center of Cold Spring Harbor for providing free DNA sequencing services and CLUSTAL W software on their website.