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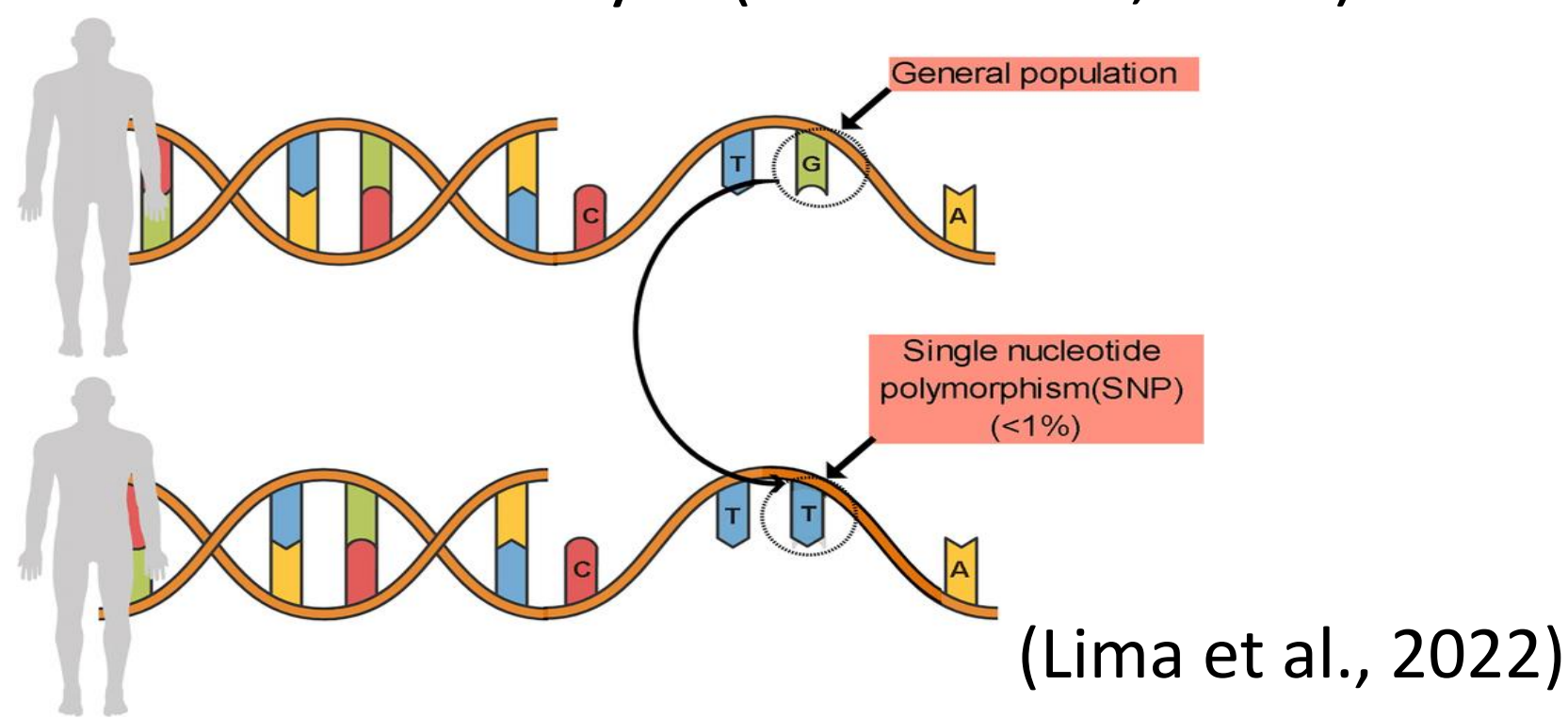
Analysis of Phenotypic Expression Associated with Different Genotypes of the Blue/Brown Eyes Single Nucleotide Polymorphism

Jade Riddle and Taylor McKinney

BIOL 250 – Introduction to Genetics and Cell Biology, Longwood University

Background

- SNP = Single-Nucleotide Polymorphism (Leisching, 2022)
 - Most frequently occurring genetic variant in the human genome
 - Helps track disease-associated genetic variants within families (Kim & Misra, 2007)
 - Predicts individual's response to drugs, environmental factors
 - Associated with cancer study (Anon, 2022)
- Human eye color
 - Under the control of approximately 16 genes
 - OCA2 considered one of the most important determinants (Eiberg et al., 2008)
 - rs12913932 region of HERC2 (SNP) regulates OCA2 expression (Sturm et al., 2007)
 - Important determinant for blue vs. brown eyes (Sturm et al., 2007)

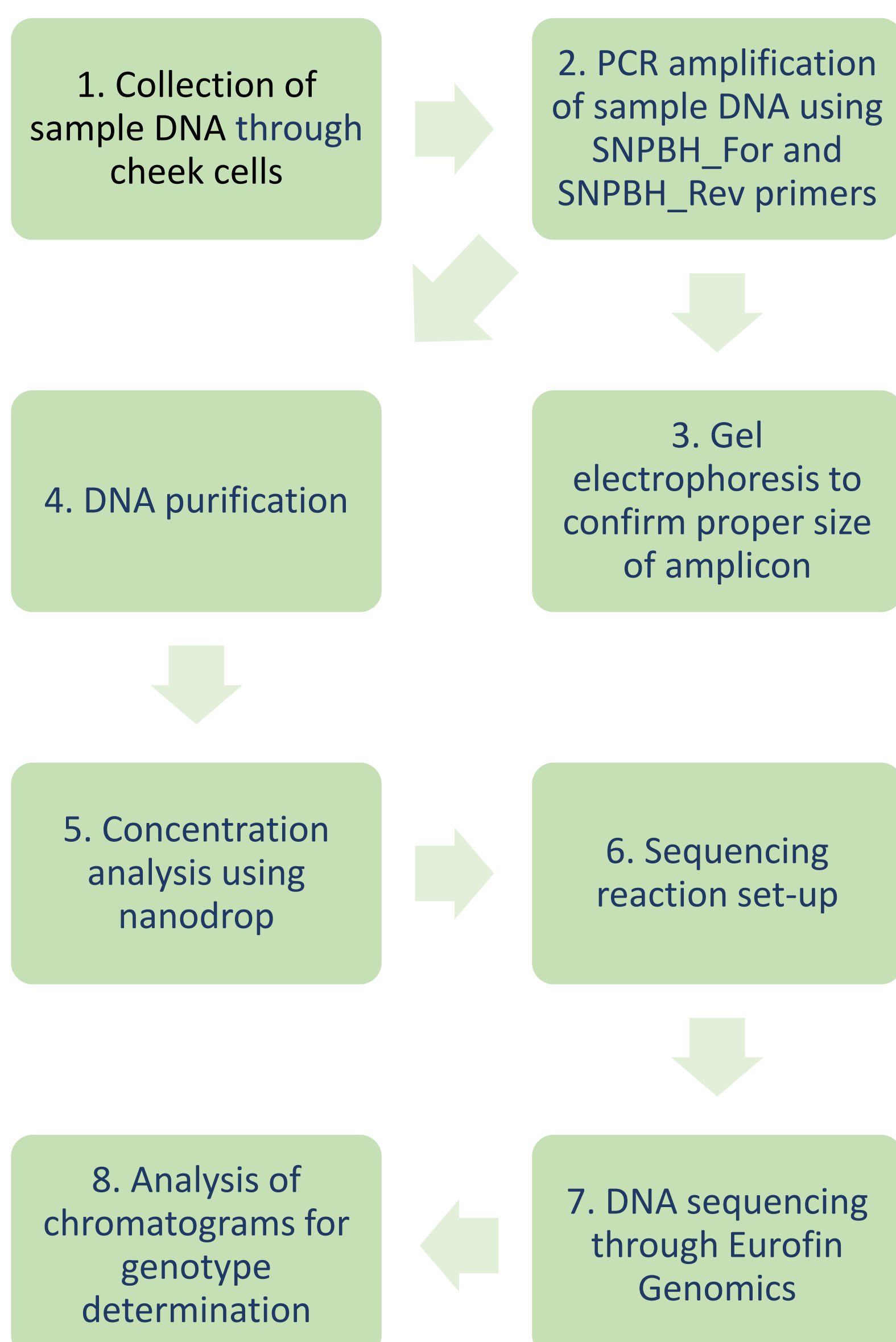


Specific Aim

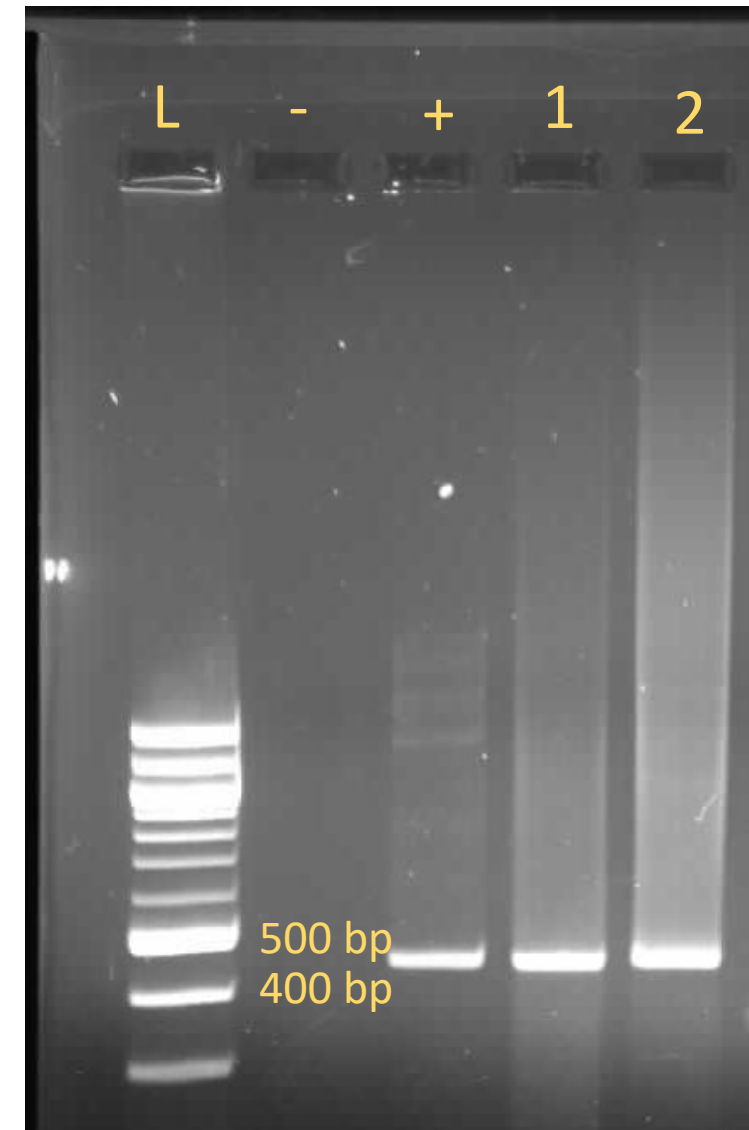
Research Question: Does the phenotypic expression of the two individuals match the expected phenotype from their DNA sequence?

Hypothesis: It was predicted that Individual 1 (Jade) would have the genotype GG due to her light-colored eyes while Individual 2 (Taylor) would have genotype AG due to her hazel eyes.

Methods



Results



Key
L - 100bp DNA Ladder
- Negative Control
+ Positive Control
1 – Jade
2 - Taylor

Figure 1: Gel electrophoresis on sample DNA. The fragments of both samples are between 400bp and 500bp at the same position as the positive control, confirming the presence of DNA and the correct size of the amplicons

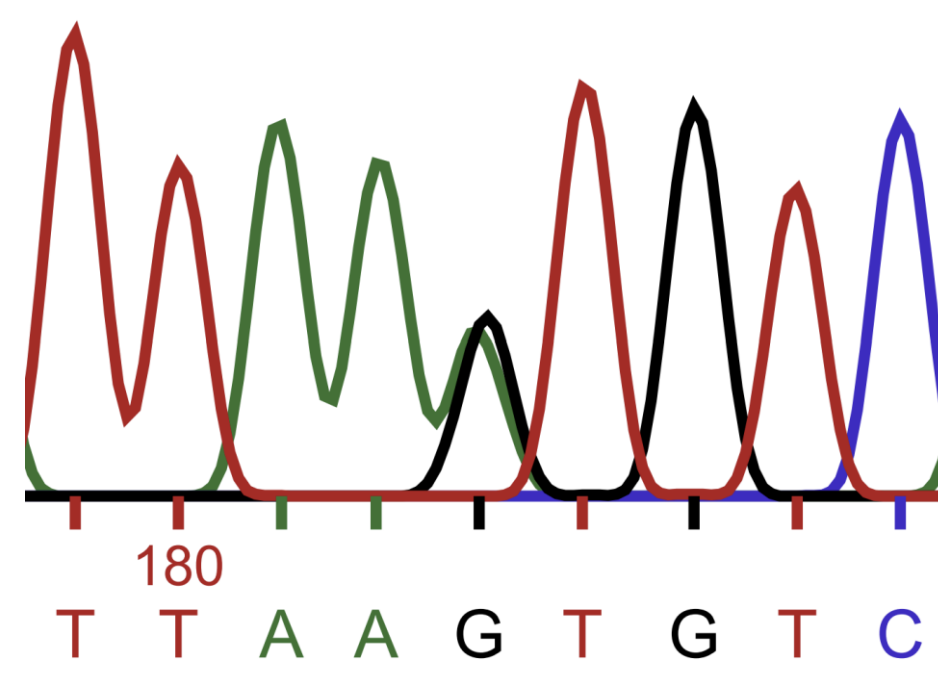


Figure 2: DNA chromatogram of individual 1. At nucleotide 183, there are two peaks present, indicating that the individual is heterozygous for this trait.

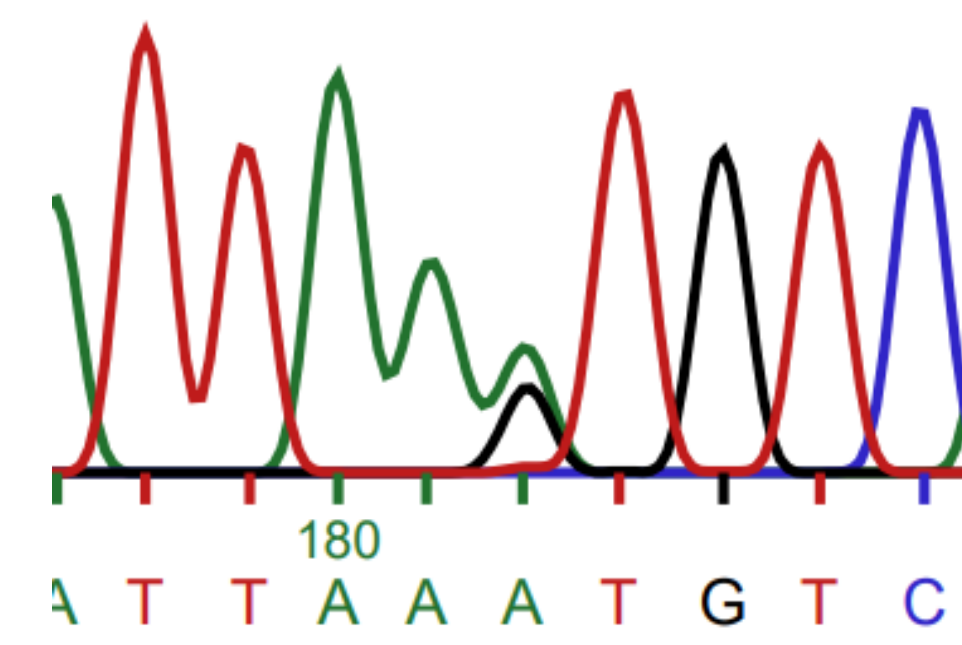


Figure 3: DNA chromatogram of individual 2. At nucleotide 182, there are two peaks present, indicating that the individual is heterozygous for this trait.

Sample	Concentration (ng/μL)	A260/A280
1	67.3	1.99
2	59.2	2.02

Table 1: Nanodrop analysis of DNA samples. The nucleic acid concentrations were found to be sufficient for use in sequencing reactions for both samples, and the A260/A280 ratio showed that both samples were thoroughly purified.

```
TGTGGGGGGGGGAAGGATTCAAGGTGCCCCAAGTATAACTC
TGAAAACATTTCTAGTCTTGTAAATCAACATCAGGGTAAAAATCATG
TGTTAATACAAAGGTACAGGAACAAAGAATTTGTTCTTCATGGCTC
TCTGTGTCTGATCCAAGAGGCGAGGCCAGTTTCATTTGAGCATTAA
GTGTCAAGTTCTGCACGCTATCATCATCAGGGGCCGAGGCTTCTCT
TTGTTTTAATTAATTGTTTTAACTGTGAGTTTATATACACTTGAAG
CAGTATACATTTAGAAATGGTCTACTTGTGTTTTCTTTGATTACTACC
CATGAGACAGTATTAGTAATTCTGGCCTATGAAATTGGCAAAGAAA
ACTACCAGTGGTGGGGAGGGTGTGAGGATGGTGGGAACA
```

Figure 4: FASTA sequence of individual 1. The SNP was located at nucleotide 183 and was determined by the analyzer to be guanine. This finding is highlighted in yellow.

```
GTTGGGGGGGGGAATGAATTCAAATGCCCCAAGTATAACTCTG
AAAACATTTCTAGTCTTGTAAATCAACATCAGGGTAAAAATCATGTGT
TAATACAAAGGTACAGGAACAAAGAATTTGTTCTTCATGGCTCTCT
GTGTCTGATCCAAGAGGCGAGGCCAGTTTCATTTGAGCATTAAAT
GTCAAGTTCTGCACGCTATCATCATCAGGGGCCGAGGCTTCTCTTT
GTTTTAATTAATTGTTTTAACTGTGAGTTTATATACACTTGAAGCA
GTATACATTTAGAAATGGTCTACTTGTGTTTTCTTTGATTACTACCA
TGAGACAGTATTAGTAATTCTGGCCTATGAAATTGGCAAAGAAAAC
TACCAGTGGTGGGGAGGGTGTGAGGATGGTGGGAACAGNAA
```

Figure 5: FASTA sequence of individual 2. The SNP was located at nucleotide 182 and was determined by the analyzer to be adenine. This finding is highlighted in yellow.

Conclusions

- Hypothesis was partially rejected
 - Both DNA samples were heterozygous
 - Genotype was not necessarily indicative of phenotype
 - Partial role of HRC2 gene in expression (Eiberg et al., 2008)
- Limitations
 - Small sample
 - Ambiguity in eye color
- Future research
 - Sequencing of close family members -> pedigree creation
 - Study of other SNPs for eye color on OCA2 gene (White & Rabago-Smith, 2011)

References

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