

Assignment 2

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Task 1

As a result of including a quality filter about 70% of the original results remain, 98 out of 138. Furthermore, the results are of a higher quality as the lower quality bases are not included in the average quality score of that position. The average quality score increases from 29.352 to 35.1487 due to the addition of the quality filter.

Task 4

The corresponding bases on the forward strand of rs12913832 and rs1129038 are TC and GA respectively. The haplotype is therefore either AC/GT or AT/GC. As my phaser doesn't manage to phase the whole file (from rs1129038 to rs12913832) I can't tell which is the correct haplotype. However as the AT/GC heterotype doesn't appear in the tables I assumed that AC/GT is correct. From the following table, where the first two haplotypes are the rs1129038 and the rs12913832 haplotypes it is calculated that there is a 0.07% chance of the AC/GT haplotype resulting in blue eyes (1+10/147) and a 92.5% chance of brown eyes (47+81+8/147). Therefore, it can be concluded that there is a much greater chance of this haplotype resulting in brown eyes [1].

Phased	Blue	Brown
ACGA/GTGA	1	47
ACGA/GTAG	10	81
ACGA/GTGG	0	8

References

- [1] Sturm, Richard A. et al. *A Single SNP in an Evolutionary Conserved Region within Intron 86 of the HERC2 Gene Determines Human Blue-Brown Eye Color*. The American Journal of Human Genetics , Volume 82 , Issue 2 , 424 - 431.