

## Sickle Cell Alleles Simbio Answers

Robust Methods for Estimating Allele Frequencies

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the distribution of allele frequencies has been a major focus in population genetics classical approaches using stochastic arguments depend highly on the choice of mutation model unfortunately it is hard to justify which mutation model is suitable for a particular sample we propose two methods to estimate allele frequencies especially for rare alleles without assuming a mutation model the first method achieves its goal through two steps first it estimates the number of alleles in a population using a sample coverage method and then models ranked frequencies for these alleles using the stretched exponential weibull distribution simulation studies have shown that both steps are robust to different mutation models the second method uses a bayesian approach to estimate both the number of alleles and their frequencies simultaneously by assuming an uninformative prior distribution the bayesian approach is also robust to mutation models questions concerning the probability of finding a new allele and the possible highest or lowest probability for a new found allele can be answered by both methods the advantages of our approaches include robustness to mutation model and ability to be easily extended to genotypic haploid and protein structure data

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