

# 1 Genotype Likelihoods from Reads

$$\begin{aligned}
L(g = 1) &= p(d | g = 1) \\
&= p(d, g = ra | g = 1) + p(d, g = rb | g = 1) + p(d, g = rc | g = 1) \\
&= \sum_a p(d | g = ra) \times p(g = ra | g = 1)
\end{aligned}$$

Here we do not favour any heterozygous genotype, and all have likelihood 1/3. This may be changed to reflect empirical or dbSNP data

$$\begin{aligned}
L(g = 1) &= 1/3 \sum_a p(d | g = ra) \\
&= 1/3 \sum_a p(d | g = ra, \text{ado}) \times p(\text{ado}) + p(d | g = ra, \text{no ado})(1 - p(\text{ado})) \\
p(d | g = ra, \text{ado}) &= p(d | g = ra, \text{drop r}) * p(\text{drop r}) + p(d | g = ra, \text{drop a}) * p(\text{drop a})
\end{aligned}$$

Here we assume either allele is equally likely to be dropped in an ado event and  $p(\text{drop r}) = p(\text{drop a}) = 0.5$ . This is unlikely to change.

$$\begin{aligned}
p(d | g = ra, \text{ado}, \text{drop a}) &= \prod_i p(d_i | g = rr) \\
p(d | g = ra, \text{ado}, \text{drop r}) &= \prod_i p(d_i | g = aa)
\end{aligned}$$

## 2 Cell-locus Posterior probabilities

Using Bayes' rule:

$$p(g = k | d) = \frac{p(d | g = k) p(g = k)}{p(d)}$$

For a cell at a given locus  $p(d)$  will be constant, therefore instead of finding it we can simply normalise:

$$p(g = k | d) = \frac{\tilde{p}(g = k | d)}{\sum_l \tilde{p}(g = l | d)}$$