Homework2

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Calculate Posterior Porbability

Define variables just as what we did in the lecture: Let $\mathbf{D} = \{\mathbf{D_1}, \mathbf{D_2}, \cdots \mathbf{D_n}\}$ be observed genotypes ($\mathbf{D_i} \perp\!\!\!\perp \mathbf{D_j}$, $\forall i \neq j$)on one specific genomic position from each reads passing that specific location. Let $\mathbf{R}$ be the reference genotype on this very same position, and $\mathbf{G}$ be underlined true genotype at this same location. Using the above settings, what we are interested in can be expressed as $P(\mathbf{G}|\mathbf{D}, \mathbf{R})$. Based on Baye's Rule, this can be rephrased as the following:

$$
\begin{aligned}
P(\mathbf{G}|\mathbf{D}, \mathbf{R}|) &= \frac{P(\mathbf{D}|\mathbf{G}, \mathbf{R}) * P(\mathbf{G}|\mathbf{R}))}{P(\mathbf{D}|\mathbf{R})} \\
&= \frac{P(\mathbf{D}|\mathbf{G}) * P(\mathbf{G}|\mathbf{R})}{P(\mathbf{D}|\mathbf{R})}
\end{aligned}
$$

given that $\mathbf{D}$ and $\mathbf{R}$ are conditional independent on $\mathbf{G}$.
Now, for each postion we want to calculate the probability of the true genotye ($\mathbf{G}$) being xx (0 copies of reference allele), xy (1 reference allele), and yy (2 reference allele) given the data.

1. To calculate the first case where $P(\mathbf{G} = xx|\mathbf{D}, \mathbf{R} = \mathbf{y}) = \frac{P(\mathbf{D}|\mathbf{G}=xx)*P(\mathbf{G}=xx|\mathbf{R}=\mathbf{y})}{P(\mathbf{D}|\mathbf{R}=\mathbf{y})}$. These three parts can be calculated separately as below:

   - $$
     \begin{aligned}
     P(\mathbf{D}|\mathbf{G} = xx) &= \prod_{i=1}^{n} P(D_i|\mathbf{G} = xx) \\
     &= \prod_{\{i|D_i = x\}} P(D_i|\mathbf{G} = xx) * \prod_{\{i|D_i \neq x\}} P(D_i|\mathbf{G} = \mathbf{xx})
     \end{aligned}
     $$

     The first probability is simply $(1 - \epsilon)$ where as the second probability is ϵ, assuming ϵ being the error made when genotyping on the read.
   - $P(\mathbf{G} = \mathbf{xx}|\mathbf{R} = \mathbf{y})$ is simply the frequency of non-reference allele squared ($(1-\rho)^2$) based on Hardy-Weinberg Equilibirum (HWE).
   - $P(\mathbf{D}|\mathbf{R} = \mathbf{y})$: This is a scaling constant and does not depend on $\mathbf{G}$, we noted it as c for now. This can be solved (by computer of course) using the fact that $P(\mathbf{G} = xx|\mathbf{D}, \mathbf{R} = \mathbf{y}) + P(\mathbf{G} = xy|\mathbf{D}, \mathbf{R} = \mathbf{y}) + P(\mathbf{G} = yy|\mathbf{D}, \mathbf{R} = \mathbf{y})$

   To sum up, the probability that $P(\mathbf{G} = xx|\mathbf{D}, \mathbf{R} = \mathbf{y}) = \frac{(1-\rho)^2}{c} \prod_{\{i|D_i = x\}}(1 - \epsilon) \prod_{\{i|D_i \neq x\}} \epsilon$.

2. $P(\mathbf{G} = xy|\mathbf{D}, \mathbf{R} = \mathbf{y}) = \frac{P(\mathbf{D}|\mathbf{G}=xy)*P(\mathbf{G}=xy|\mathbf{R}=\mathbf{y})}{P(\mathbf{D}|\mathbf{R}=\mathbf{y})}$

   - $$
     \begin{aligned}
     P(\mathbf{D}|\mathbf{G} = xy) &= \prod_{i=1}^{n} P(D_i|\mathbf{G} = xy) \\
     &= \prod_{i|D_i \in x,y} P(D_i|\mathbf{G} = xy) * \prod_{i|D_i \notin x,y} P(D_i|\mathbf{G} = xy)
     \end{aligned}
     $$

     The first probability is $\frac{1}{2}(1 - \epsilon) + \frac{\epsilon/3}{2}$ and the second probability is $\frac{\epsilon}{3}$.

   - $P(\mathbf{G} = \mathbf{xy}|\mathbf{R} = \mathbf{y})$ is simply $2 * \rho * (1 - \rho)$ based on HWE.
   - $P(\mathbf{D}|\mathbf{R} = \mathbf{y})$: as before, this is a constant c.

   To sum up, the probability that $P(\mathbf{G} = xy|\mathbf{D}, \mathbf{R} = \mathbf{y}) = \frac{2*\rho*(1-\rho)}{c} \prod_{\{i|D_i \in x,y\}} \frac{1-2/3\epsilon}{2} * \prod_{\{i|D_i \notin x,y\}} \frac{\epsilon}{3}$

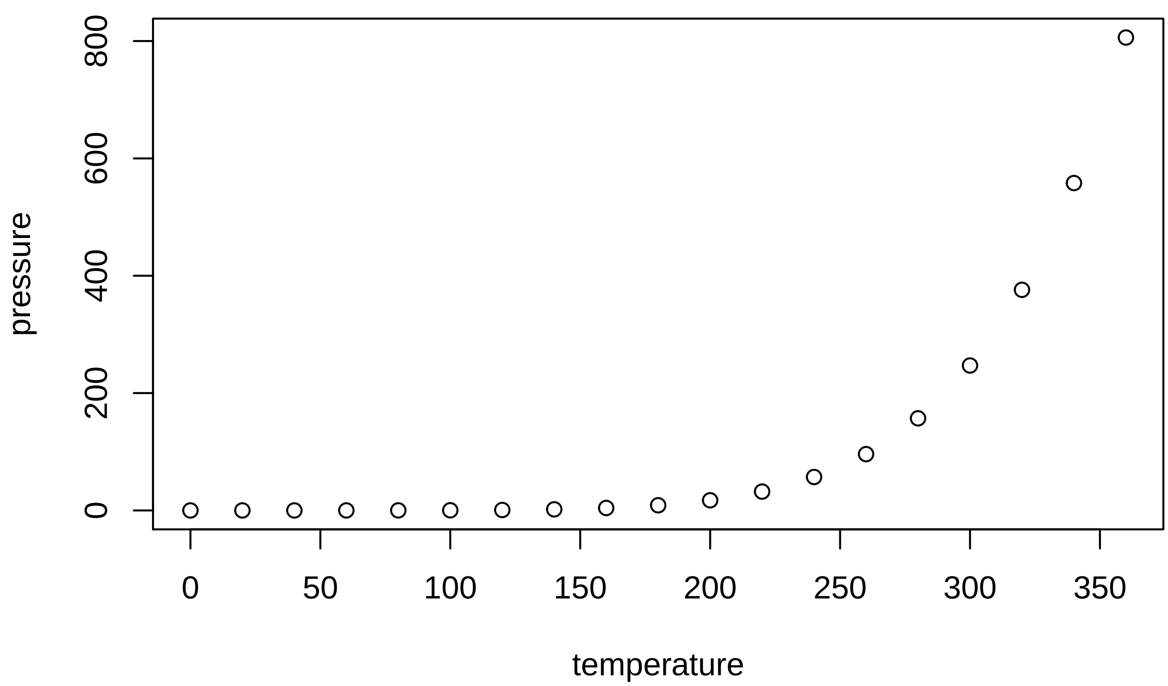
3. The last probability $P(\mathbf{G} = yy|\mathbf{D}, \mathbf{R} = \mathbf{y})$ is very similar to the first case except that $P(\mathbf{G} = \mathbf{yy}|\mathbf{R} = \mathbf{y}) = \rho^2$, and thus the probability is $P(\mathbf{G} = yy|\mathbf{D}, \mathbf{R} = \mathbf{y}) = \frac{\rho^2}{c} \prod_{\{i|D_i = x\}} (1 - \epsilon) \prod_{\{i|D_i \neq x\}} \epsilon$.

```r
summary(cars)
```

```
##      speed           dist       
##  Min.   : 4.0   Min.   :  2.00  
##  1st Qu.:12.0   1st Qu.: 26.00  
##  Median :15.0   Median : 36.00  
##  Mean   :15.4   Mean   : 42.98  
##  3rd Qu.:19.0   3rd Qu.: 56.00  
##  Max.   :25.0   Max.   :120.00  
```

Including Plots

You can also embed plots, for example:



Note that the `echo = FALSE` parameter was added to the code chunk to prevent printing of the R code that generated the plot.