

N0 and N1 calculation

N0 and N1 Calculations

Let $X = (X_1, X_2, \dots, X_m)$ be the information on the number of copies of m cSNV, e.g. $X = (0, 0, 1)$ or $X = (0, 1, 1)$ for $m = 3$.

m = number of cSNV's in the model.

$X_j \in 0, 1, 2$ for $j = 1, 2, \dots, m$

Logistic model to measure the effect of number of copies of cSNV on the probability of having disease:

$$\text{logit}(P(D|X)) = \beta_0 + \beta_1 \sum_{i=1}^m X_i$$

$P(D)$ is the probability of disease in the population of 3100 individuals.

$$P(D) = \int P(D|X)P(X)dX = E[P(D|X)] = \frac{1}{N} \sum_{j=1}^N P(D|X_j)$$

where $P(D|X_j)$ is the probability of disease for the j -th individual. Let us assume each person carries zero or one cSNV, then:

$$\text{if } \sum_{i=1}^m X_i = 0 \text{ we have, } \text{logit}P(D|X) = \frac{e^{\beta_0}}{1+e^{\beta_0}}$$

$$\text{if } \sum_{i=1}^m X_i = 1 \text{ we have, } \text{logit}P(D|X) = \frac{e^{\beta_0+\beta_1}}{1+e^{\beta_0+\beta_1}}$$

and we can write $P(D)$ in the following form:

$$P(D) = pd = \frac{1}{N} \sum_{j=1}^N P(D|X_j) = \frac{1}{N} (N_0 \times a + N_1 \times b)$$

where in this formula:

$$a = \frac{e^{\beta_0}}{1+e^{\beta_0}}$$

$$b = \frac{e^{\beta_0+\beta_1}}{1+e^{\beta_0+\beta_1}}$$

As $N_0 + N_1 = 3100$ we can calculate N_1 as follows:

$$N_1 = \frac{3100 \times (pd - a)}{b - a}$$

A function to calculate N_1 for different values of pd :

```
N1_Calc = function(pd){  
  
  beta0 = -10  
  beta1 = 16  
  
  a = exp(beta0) / ( 1 + exp(beta0) )  
  b = exp(beta0+beta1) / ( 1 + exp(beta0+beta1) )  
  
  res = ( 3100 * (pd - a) ) / (b - a)  
  return(res)  
}
```

The N_1 value if the probability of disease in the population is 0.05:

```
round( N1_Calc(0.05) )
```

```
## [1] 155
```