

2018 SISG Practical

Session #8: Prediction

1. For the disease(s) you have been evaluating in the even numbered modules, now consider the column titled 'genotypes'. These are the observed genotypes for one individual.
2. Calculate the polygenic risk score for these genotypes and your disease of interest. Genotypes can be coded as 0, 1, or 2 risk alleles. Then the risk score is calculated as

$$s_j = \frac{\sum_{i=1}^m \beta_i x_{ij}}{m}$$

where the beta are the given log(OR) estimates, and x are the {0,1,2} genotype codings. Note that the score doesn't necessarily need to be divided by m.

3. Interpret this result. How can you compare this finding to the average risk in the population? Hint: recall how we calculated the mean risk in previous modules, assuming Hardy-Weinberg equilibrium
$$M = (1-p)^2 + 2p(1-p)OR + p^2 OR^2$$
4. How would you counsel the person with these genotypes regarding their disease risk?