

2018 SISG Practical

Session #4: Measures of variation

1. Use the same dataset and your two selected diseases from the Session #2 Practical.
2. For each disease, extract all risk allele frequencies and summary statistics of association (i.e., ORs). See below screenshot for input data required. Note that we will be estimating the heritability of liability.
3. Look online for an estimate of the lifetime disease risk (or approximate with prevalence) and lambda sibling.
4. Use these data to calculate different measures of variation explained using the tool at: <http://cnsgenomics.com/shiny/INDI-V/>
5. Interpret output with regard to differences in the measures and how much genetic variation has been explained.

Input Variables

	Risk variant	Name	e.g. rs number
1	RAF	RAF	Risk allele frequency (risk allele = B).
2	RRBb	RR_{Bb}	Genotype relative risk of heterozygote Bb relative to homozygote bb.
3	RRBB	RR_{BB}	Genotype relative risk of homozygote BB relative to homozygote bb (default multiplicative $RR_{BB} = RR_{Bb}^2$).
4	K	K	Baseline population risk of disease.
5	lambda_sib	λ_s	Sibling relative risk (>1): Increased risk of disease in siblings of those affected- used with K to estimate heritability of liability - OR:
6	h2L_total	h_L^2	Heritability of liability (0-1): The proportion of variance in phenotypic liability attributed to additive genetic effects - estimated from twin/family studies - used with K to estimate sibling recurrence risk.