SISG 2022: Module 11
Session 1: Introduction

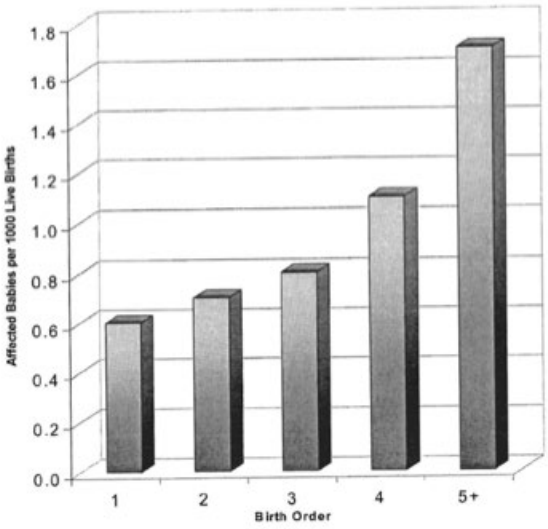
1. Each person, please introduce yourself to the other members in your group:
    1. Your name and pronouns. Your position (student, researcher) and affiliation (what University or institute).
    2. What are your strengths in your training so far? (e.g., is your background in genetics, biostatistics, law?)
    3. What prompted you to take this course? What are you hoping to learn?

2. Once everyone is introduced, discuss in your group:
    1. Why do we study the role of genetics in human disease?

SISG 2022: Module 11
Session 2: Introduction to Genetic Epidemiology

1. The following questions are based on this figure:

Confounding example: Birth order and Down syndrome



1. **Can you think of a factor that would confound the observed association?**
2. **How can you use data on your proposed confounding factor to reassess the association between birth order and Down Syndrome?**
3. **Can you think of potential confounders in genetic epidemiology?**

Data from Stark and Mantel (1966)

Source: Rothman 2002

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Session 3: Human Genetic Variation

1. A recent study sequenced the genome of 2,504 individuals and identified 84.7 million SNPs (single nucleotide polymorphisms) between all participants. On average, each individual carried 3.5-4.3 million SNPs each. About 0.5% of those SNPs were in coding regions of genes. Remember, 1.5% of the genome is in a coding region. Why might only 0.5% of variants be in coding regions compared to what would be expected if SNPs were randomly allocated throughout the genome?

2. Match the genetic term with the definition:

   a. Nonsense
   b. Heterozygous
   c. Exon
   d. Allele
   e. Synonymous
   f. Missense
   g. Non-coding region
   h. Haplotype
   i. Autosomal
   j. Phenotype
   k. Genotype
   l. Frameshift
   m. Intron
   n. Homozygous

___ Alternative forms of a gene or DNA base.
___ Genetic makeup of an individual at a particular DNA location based on both alleles.
___ Genotype consisting of two different alleles at a particular location.
___ DNA base change that does not change the translated amino acid.
___ Genotype consisting of two of the same alleles at a particular location.
___ Observable characteristics resulting from a genotype.
___ Concerning the 22 pairs of chromosomes that are not sex chromosomes.
___ Portion of gene that does not code for amino acids and appears in between exons.
___ Insertion or deletion mutation that changes the whole subsequent sequence of amino acids by changing the 3-codon groups for generating amino acids.
___ Portion of gene that encodes amino acids.
___ Section of DNA that does not become protein.
___ Substitution of a single DNA base that causes a stop in protein production.
___ DNA base change that changes the translated amino acid.
___ Set of DNA variations at several positions that are inherited together.

3. Look up “rs7412” in dbSNP (https://www.ncbi.nlm.nih.gov/snp/).
   a. What are the DNA bases identified at this location?
   b. What gene is this SNP located in?
   c. What is the effect of this SNP on the amino acid sequence?

Click on the “frequency” tab to the left. What is the frequency of the minor allele (less common allele) in the 1000 Genomes study overall? How do these frequencies differ by ancestral subgroup within this study?