# Kahoot

**How many chromosomes does an average human have?**

A) 22

B) 23

C) 44

D) 46

**Which of the following is a type of genetic variation?**

A) insertions/deletions

B) chromosomal structural variants

C) single nucleotide polymorphisms  
D) all of the above

**Polymorphisms...**

A) may be single nucleotide variants or structura variants

B) are rare

C) can be found in just a few cells in the body  
D) are only present in germline cells

**What is true about genotypes and phenotypes?**

A) All phenotypes can be fully predicted from genotypes

B) The phenotype describes what your DNA looks like

C) We need information from both chromosomes in a pair to know the genotype  
D) Genotypes are different types of genes

**Heterozygous = 2 copies of the same nucleotide base on the same strand**

TRUE/FALSE

**Confounders are associated with both the exposure and the outcome**

TRUE/FALSE

**What is true about the HWE?**

A) Real populations are always in HWE

B) Under HWE, the allele frequencies and genotype frequencies are identical

C) Under HWE, the genotype frequencies can be calculated from allele frequencies  
D) Genotype frequencies will stay the same even if allele frequencies change

**Two genetic variants are in LD if they are...**

A) on the same chromosome

B) in the same gene

C) transmitted together from a parent to a child  
D) transmitted together more often than expected by chance

**Heritability quantifies...**

A) the contribution of genetic variation to the variability of a trait

B) how important genes are for the absolute value of the trait

C) the value of a quantitative trait  
D) the similarity of twin pairs

**What is true about IBS and IBD?**

A) IBS segments are always also IBD

B) An IBD segment in two individuals have the same ancestral origin

C) Individuals with IBD segments must have at least one parent in common  
D) Relatedness can be inferred from any IBS segment

# The Hardy-Weinberg Equilibrium (HWE)

1. A SNP has the following observed genotype frequencies:

|  |  |  |
| --- | --- | --- |
| **AA** | **AT** | **TT** |
| 0.400 | 0.463 | 0.137 |

What are the allele frequencies under HWE?

(A)

(T)

1. In a population of 300 individuals, we observe the following distribution of the SNP above. What is the expected genotype distribution under HWE?

|  |  |  |  |
| --- | --- | --- | --- |
|  | AA | AT | TT |
| Observed distribution | 120 | 139 | 41 |
| Expected distribution | 119.07 | 139.86 | 41.07 |

AA: 300 x 0.632 = 119.07

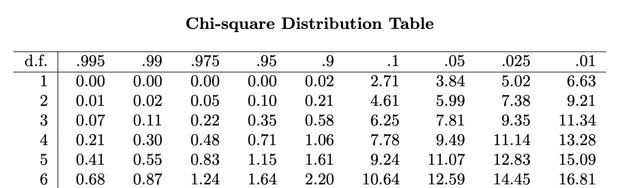
AT: 300 x 2 x 0.63 x 0.37 = 139.86

TT: 300 x 0.372 = 41.07

1. Calculate the test statistic for the above SNP. Is there evidence that the SNP is in HWE?

A picture containing text, watch

Description automatically generated xi = observed count, mi = expected count



Deviations between the expected and observed genotype frequencies:

AA: (120-119.07) = 0.93

AT: (139-139.86) = -0.86

TT: (41-41.07) = -0.07

c2 = (0.932/119.07) + (0.862/139.86) + (0.072/41.07) = 0.0073 + 0.0053 + 0.00012

= 0.0127

1. Is there statistical evidence that the marker is in HWE?

Yes, there is evidence that the SNP is in HWE because the c2 < 5.99 so we therefore keep the null hypothesis and conclude that there is no significant difference between the observed and expected genotype distributions.

# Linkage Disequilibrium:

Consider two loci on the same chromosome: Locus 1 (alleles A and a) and locus 2 (alleles B and b). In 1000 chromosomes (500 individuals), we observe the following number of individuals with the respective haplotypes:

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
|  | AB | Ab | aB | ab |
| Observed number | 450 | 50 | 100 | 400 |

1. What are the allele frequencies for each of the four alleles?

A: (450 + 50) / 1000 = 0.5,

a: (100 + 400) / 1000 = 0.5

B: (450 + 100) / 1000 = 0.55

b: (50 + 400)/ 1000 = 0.45

1. What are the expected haplotype frequencies if the two loci are in linkage equilibrium?

AB: 0.5 x 0.55 = 0.275

Ab: 0.5 x 0.45 = 0.225

aB: 0.5 x 0.55 = 0.275

ab: 0.5 x 0.45 = 0.225

1. Calculate the LD coefficient D and the standardized D’ for the alleles A and B. Are these alleles in LD?

Observed haplotype AB frequency = 450/1000 = 0.45

Expected haplotype AB frequency (product of individual allele frequencies) = 0.275

D = 0.45 - 0.275 = 0.175

Dmax = the smaller value of 0.5\*0.45 and 0.5\*0.55 because D>0.

D’ = D/Dmax = 0.175/0.225 = 0.77

D’>0, so the alleles are in LD.