

Coursenote of:
MAST10016 - Mathematics for Biomedicine

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Chapter 1

Population genetics

1.1 Basic terminology in genetics

- Locus: The position of a gene in a particular chromosome
- Autosomes: Homologous pairs of genes
- Sex chromosomes: Non homologous pairs of genes which determines the sex

1.2 Co-dominance and incomplete dominance

- Co-dominance: The contribution of both different alleles A and a are visible in the phenotype
- Incomplete dominance: The phenotype of the heterozygous genotype is intermediate to the phenotype of the homozygous genotypes

1.3 Sex chromosomes

If A and a are alleles of an **X-linked gene**, then the genotypes are

- Female: AA, Aa, aa
- Male¹ A-, a-

1.4 Inheritance

- **Gametes** are produced in the testes/ovaries by meiosis thus haploid
- Haploid sperm and ova unite at fertilization to form a **zygote**, which is diploid

1.5 Segregation principle

- Genes randomly receive one of the adult's two alleles for each autosomal gene with equal probability
- Gametes randomly receive one of the adult's two sex chromosomes with equal probability
- This occurs independently for genes on different chromosomes

1.6 Probability laws

1.6.1 Multiplication law for independent events

If E_1 and E_2 are independent events, then $\Pr(E_1 \cap E_2) = \Pr(E_1) \times \Pr(E_2)$.

¹These nomenclatures can also be written as A-, a- or X^AY, X^aY

1.6.2 Addition law for mutually exclusive events

If E_1 and E_2 are independent events, then $\Pr(E_1 \cup E_2) = \Pr(E_1) + \Pr(E_2)$.

1.7 Prevalence and incidence in genetic disorders

- Prevalence: The total number or proportion of the population affected by the genetic disorder at the given time
- Incidence: The number or proportion of new births that are affected by the genetic disorder

1.8 Genotype numbers and frequencies

- N : Total population size
- N^{AA} : Number of genotype AA individuals in the population
- f^{AA} : Frequency of genotype AA in the population
- Likewise for N^{Aa} , f^{Aa} , N^{aa} , etc.
- Note that $N^{AA} + N^{Aa} + N^{aa} = N$ and $f^{AA} + f^{Aa} + f^{aa} = 1$

1.9 Allele numbers and frequencies

- N^A : Number of A alleles in the population
- f^A : Frequency of A alleles in the population
- Likewise for N^a and f^a

1.10 Calculating allele frequencies from genotype frequencies

Since each AA genotypes has 2 A alleles, and each Aa genotypes has 1, we have

$$N^A = 2N^{AA} + N^{Aa} \quad N^a = 2N^{aa} + N^{Aa} \quad (1.1)$$

Each individual has 2 alleles, so the total size of the allele pool is $2N$. The frequencies are

$$f^A = \frac{N^A}{2N} \quad f^a = \frac{N^a}{2N} \quad (1.2)$$

Combine equations 1.1 and 1.2 to get

$$f^A = \frac{N^A}{2N} = \frac{2N^{AA} + N^{Aa}}{2N} = \frac{N^{AA}}{N} + \frac{1}{2} \frac{N^{Aa}}{N}$$

Therefore,

$$f^A = f^{AA} + \frac{1}{2} f^{Aa} \quad f^a = f^{aa} + \frac{1}{2} f^{Aa}$$