

Lynch & Wash Chapter 2 - Properties of distributions

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Research Interest: Genomic selection, statistical and machine learning models

Outline

- 1 Parameters of Univariate Distributions
 - Characters
 - Distributions
 - The Normal distribution
 - Truncated Normal distribution
 - Confidence intervals

Types of Characters

Types of characters studied by biologists:

- **Meristic characters** (range of discrete classes)
 - ▶ Example: Leaf numbers
- **Metric characters** (range of discrete classes)
 - ▶ Example: Plant height
- **Binary characters** (range of discrete classes)
 - ▶ Example: Survival at a fixed age

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Types of Distributions

We usually can describe data using three different types of distributions:

- Univariate distribution (UD):

Definition: **"UD describes the relative frequency of phenotypes for a single trait"**

- Bivariate distribution (BD):

Definition: **"BD describes the mutual distribution of two characters"**

- Multivariate distribution (MD):

Definition: **"MD corresponds to the joint distribution of more than two traits"**

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Types of Distributions

- **Goal of statistics:** Fit simple mathematical functions to data

- Types of probability distributions:

- ▶ Discrete: **some variable z (e. g. offspring number) is completely described by $P(z = z_i)$**

Book's example: $P(z = z_1) = \frac{15}{1005}$, given $z_1=1$, that is, proportion of mothers that produce a single offspring

- ▶ Continuous: **Point probability is infinitesimal small (we should compute interval prob.)**

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Distributions properties

■ Key property:

▶ $\int_{z_{\min}}^{z_{\max}} p(z) dz = 1$ (Continuous case)

▶ $\sum_{i=1} p(z_i) = 1$ (Discrete case)

■ What is a easy way to integrate probability distributions?

▶ **Monte Carlo Integration!**

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- ▶ **Monte Carlo Integration!**

Example of computing probabilities

Example 1: Suppose that z is a continuously distributed in the range of 0 to ∞ with pdf negative exponential distribution:

$$p(z) = \frac{1}{\lambda} \exp \left[\frac{-z}{\lambda} \right]$$

What is the probability that a random sample will have z in the range of $\frac{1}{4}$ to $\frac{1}{2}$?

$$P\left(\frac{1}{4} \leq z \leq \frac{1}{2} | \lambda\right) = \int_{\frac{1}{4}}^{\frac{1}{2}} p(z) dz = \int_{\frac{1}{4}}^{\frac{1}{2}} \frac{1}{\lambda} \exp \left[\frac{-z}{\lambda} \right] dz$$

Rule: $\int e^{ax} dx = \frac{1}{a} e^{ax}$, in our case:

$$\begin{aligned} \int_{\frac{1}{4}}^{\frac{1}{2}} p(z) dz &= \frac{1}{\lambda} - \lambda \exp \left[\frac{-z}{\lambda} \right] \Big|_{\frac{1}{4}}^{\frac{1}{2}} = -\exp \left[\frac{-z}{\lambda} \right] \Big|_{\frac{1}{4}}^{\frac{1}{2}} = \\ &= \exp \left[\frac{-1}{4\lambda} \right] - \exp \left[\frac{-1}{2\lambda} \right] \end{aligned}$$

for $\lambda = \frac{1}{2}$, then $P\left(\frac{1}{4} \leq z \leq \frac{1}{2} | \lambda\right) = 0.239$

Example of computing probabilities

Monte Carlo integration: **R code!**

Parameters vs Estimates

- True parameters are only obtained if all population units are measured with high accuracy
- Estimates are statistics obtained from analysing sampled data
- Accuracy of estimates rely on **experimental setting**, **measurement apparatus** and the **sample size**

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Parameters vs Estimates

- Most useful probability density function are defined by:

- **Central location:**

- ▶ $\mu = \int_{-\infty}^{+\infty} z p(z) dz = E(z)$

- ▶ $\mu = \sum_{i=1} p(z = z_i)$

- **Variance parameter:**

- $$\sigma^2 = \int_{-\infty}^{+\infty} (z - \mu)^2 p(z) dz = E[(z - \mu)^2]$$

- Some useful expectation properties:

- ▶ $E(x + y) = E(x) + E(y)$

- ▶ $E(c x) = c E(x)$

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Parameters vs Estimates

- Variance has the problem of being harder to estimate with small samples

Recommendation: $E [E(z - \mu)^2] = \frac{1}{n-1} \sum_{i=1} (y_i - \bar{y})^2$

- Square root of the variance: standard deviation or scale parameter
- Relative measure of dispersion: $CV(z) = \frac{SD(z)}{\bar{z}}$
- Measure of a asymmetry of a distribution (skewness):

$$\mu_3 = E [(z - \mu)^3] = \int_{-\infty}^{+\infty} (z - \mu)^3 p(z) dz$$

$$\mu_3 = E [z^3] - 3\mu E [z^2] + 2\mu^3$$

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Parameters vs Estimates

- The degree of asymmetry can also be obtained by the coefficient of skewness:

- ▶ $k_3 = \frac{Skw(z)}{Var(z)^{\frac{3}{2}}}$

- ▶ $k_3 > 0$ longer tail to the right

- ▶ $k_3 < 0$ longer tail to the left

- ▶ $k_3 = 0$ perfectly symmetrical distribution

- General expression for the r th moment about the mean:

$$\mu_r = \int_{-\infty}^{+\infty} (z - \mu)^r p(z) dz$$

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Normal p.d.f. features

- In most cases, large datasets when plotted in form of histograms often approximate bell-shaped distribution
- Normal distribution developed by DeMoivre(1738), LaPlace(1778) and **Gauss(1801)**

- Probability density function ($z(\mu, \sigma^2)$)

$$p(z) = (2\pi\sigma^2)^{-\frac{1}{2}} \exp \left[-\frac{(z-\mu)^2}{2\sigma^2} \right]$$

- **Some properties:** mean = median, symmetrical and has two parameters, the mean (location) and variance (scale)
- **Key properties:** Nice mathematical properties, random variables that deviates normality can be rescaled by transformation

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- Why most of the quantitative traits follows a normal p.d.f. ?
 - ▶ **Reason:** Central limit theorem + infinitesimal genetic architecture hypothesis
- Central limit theorem states that "the sum of a number of independent random variables approaches normality as the number of variables increases"
- **Tiny limitation:** possibility of negative values realizations (sampling)

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Normal p.d.f. features

- Discrete data with large number of categories approximate normality as these numbers increases (result of central limit theorem)
- It is frequently useful to work with the standardized form of the normal density:

$$\triangleright p(z') = (2\pi)^{-\frac{1}{2}} \exp \left[-\frac{(z')^2}{2} \right]$$

- **Other features:**

- $\mu_3 = 0$ (symmetrical distribution)
- $k_4 = \frac{Kur(z) - 3[Var(z)]^2}{[Var(z)]^2} = 0$ (index to measure kurtosis)
- Deviation from normality:** $k_4 > 0$ (leptokurtic, e. g. Cauchy density), $k_4 < 0$ (platykurtic, e. g. Uniform density)

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Truncated Normal distribution (TND) features

- The TND corresponds to some subset values of the normal distribution
- Extremely import for theory of **truncation selection**
- Truncation selection:
 - ▶ Critical phenotype T is called truncation point
 - ▶ Mean of the subset of the distribution (mean of the selected fraction of genotypes) can be computed by:

$$\mu_S = \frac{\int_T^{+\infty} zp(z) dz}{\int_T^{+\infty} p(z) dz}$$
 - ▶ After reparametrizations and the integration we got:

$$\mu_S = \mu + \frac{\sigma p_T}{\Phi_T}, \text{ where } p_T = (2\pi)^{-\frac{1}{2}} \exp \left[-\frac{(T-\mu)^2}{2\sigma^2} \right]$$
- p_T can be obtained by the *dnorm*($T,0,1$) R function

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- *Directional selection differential*: $\mu_s - \mu = \frac{\sigma p_T}{\Phi}$
- Units of phenotypic standard deviations: $\frac{(\mu_s - \mu)}{\sigma} = \frac{p_T}{\Phi_T}$
- Variance after selection: $\sigma_s^2 = \left[1 + \frac{p_T z'}{\Phi_T} - \left(\frac{p_T}{\Phi_T} \right)^2 \right] \sigma^2$
- which gives the fraction of phenotypic variance after selection $\left(\frac{\sigma_s^2}{\sigma^2} \right)$

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- Confidence intervals are obtained by theoretical mathematical expressions based upon frequentist theory:

$$\alpha = P[(\bar{z} - \Delta) \leq \mu \leq (\bar{z} + \Delta)] = \int_{\frac{-\Delta}{\sigma(\bar{z})}}^{\frac{+\Delta}{\sigma(\bar{z})}} p(z') dz'$$

in which $z' = \frac{(\bar{z} - \mu)}{\sigma(\bar{z})}$ and $\frac{\Delta}{\sigma(\bar{z})}$ distance in standard errors that the observed statistic and parametric value will lie with probability α

- Mean case: $\bar{z} \pm 1.96 \left[\frac{\text{var}(z)}{n} \right]^{\frac{1}{2}}$
- Variance case (approximation): $2\sqrt{\text{var}(z)}$ (large sample) - 95% confidence interval

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Thanks!

"les charmes enchanteurs de cette sublime science ne se décèlent dans toute leur beauté qu'à ceux qui ont le courage de l'approfondir" - Letter from Gauss to Sophie Germain (30 April 1807).