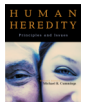
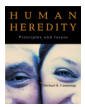
Chapter 5
Complex Patterns of Inheritance



Phenotypes Can Be Discontinuous or Continuous

- **Discontinuous variation** shows distinct phenotypes
  - Short and tall peas phenotypes

- **Continuous variation** shows a series of overlapping phenotypic classes
  - Height in humans



Continuous and Discontinuous Variation

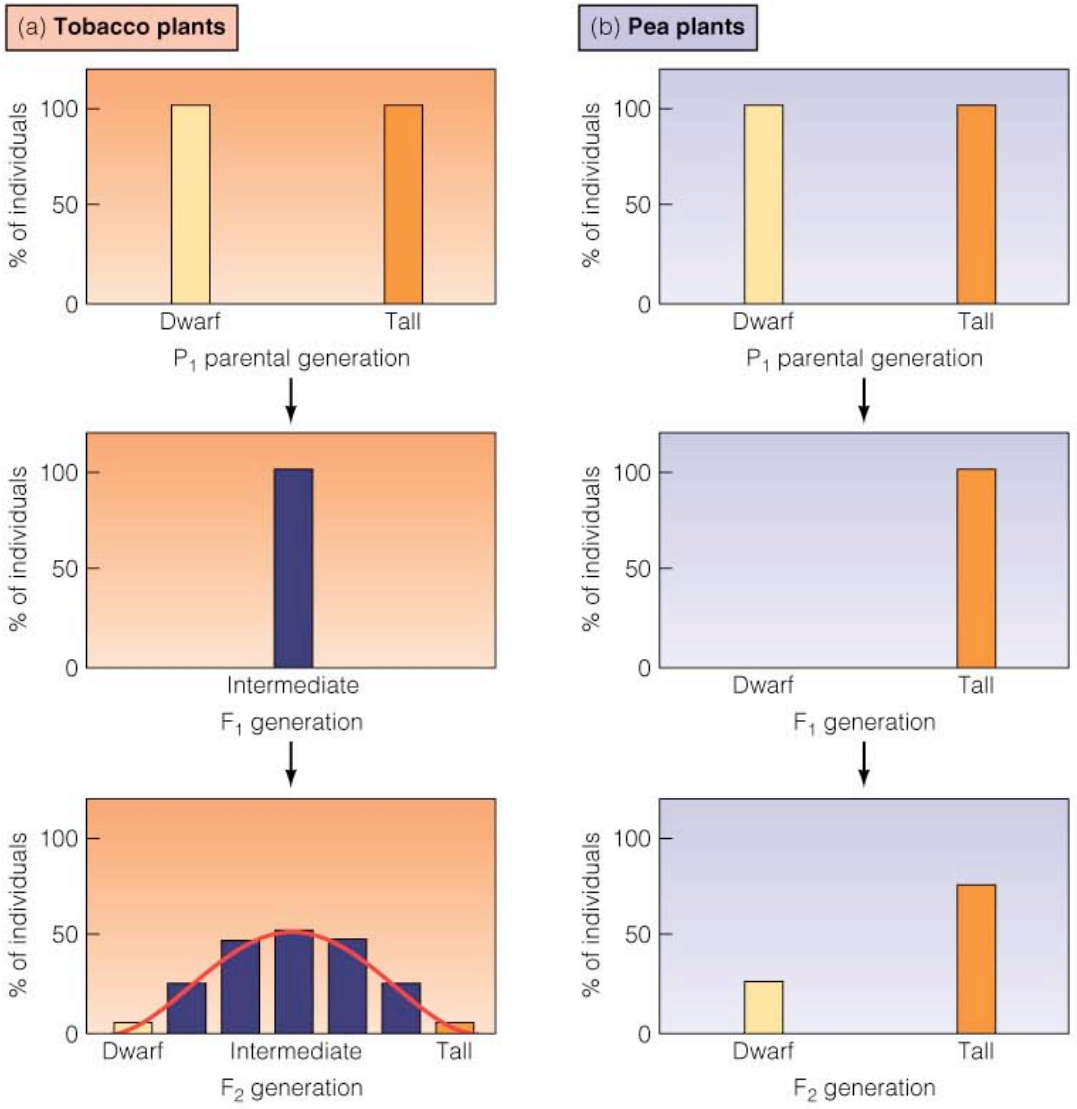
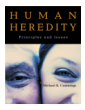


Fig. 5.2



Continuous Variation in Humans

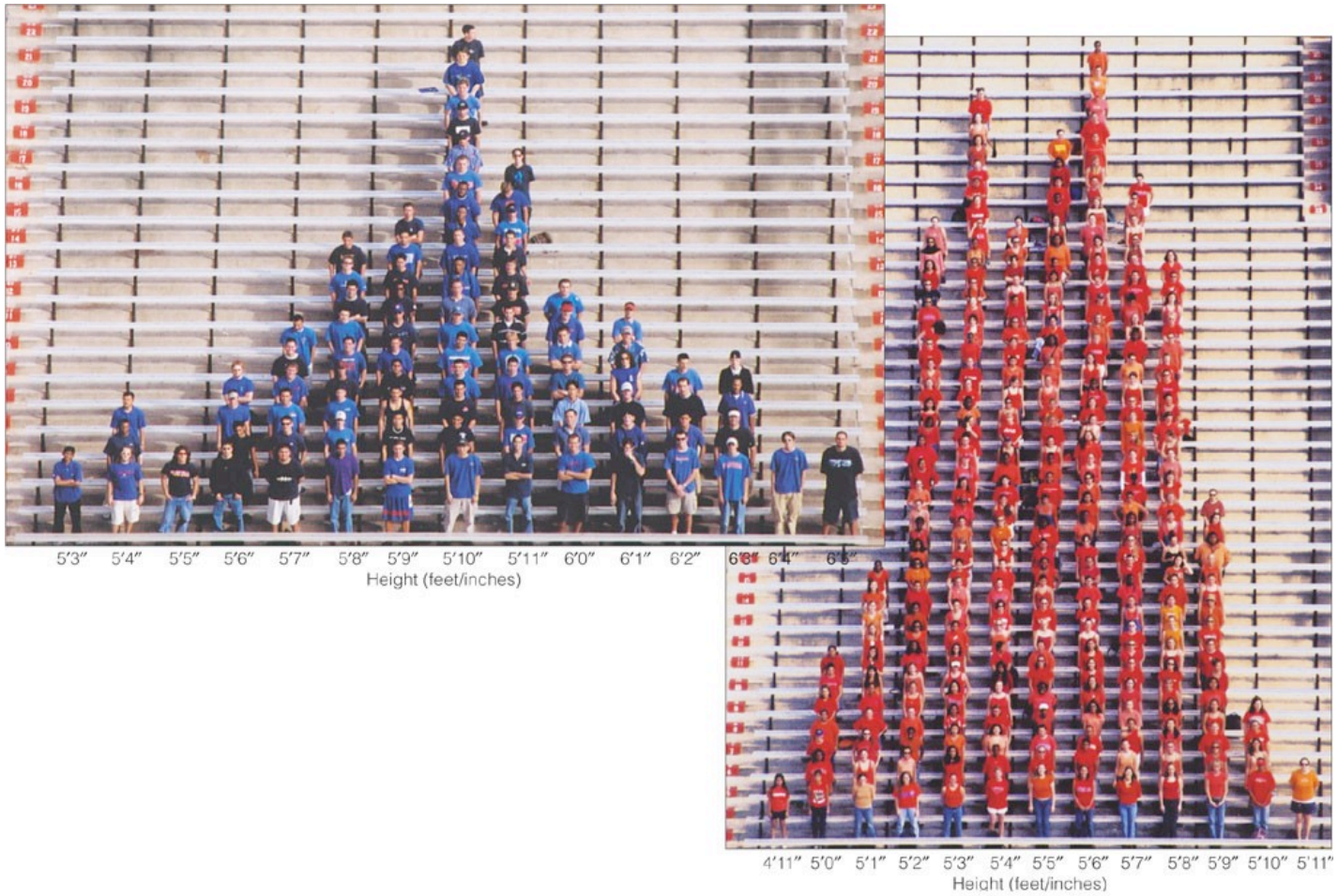
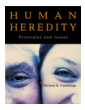


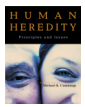
Fig. 5.1



Genotype + Environment

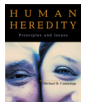
Produce the Phenotype

$P = G+E$



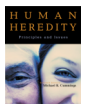
Terms

- **Polygenic traits** are determined by two or more genes
- **Multifactorial traits** are controlled by two or more genes and show significant interaction with the environment
- **Complex traits** are ones where relative contribution of genes and environment are not yet established



Polygenic Inheritance

- Traits are usually quantified by measurement
- Two or more genes contribute to the phenotype
- Phenotypic variation varies across a wide range
- Better analyzed in populations than in individuals
- Example: human eye color



- As the number of loci increases, the number of classes increases
- As classes increase, phenotypic difference between classes decreases
- Averaging out of the phenotype is called **regression to the mean**

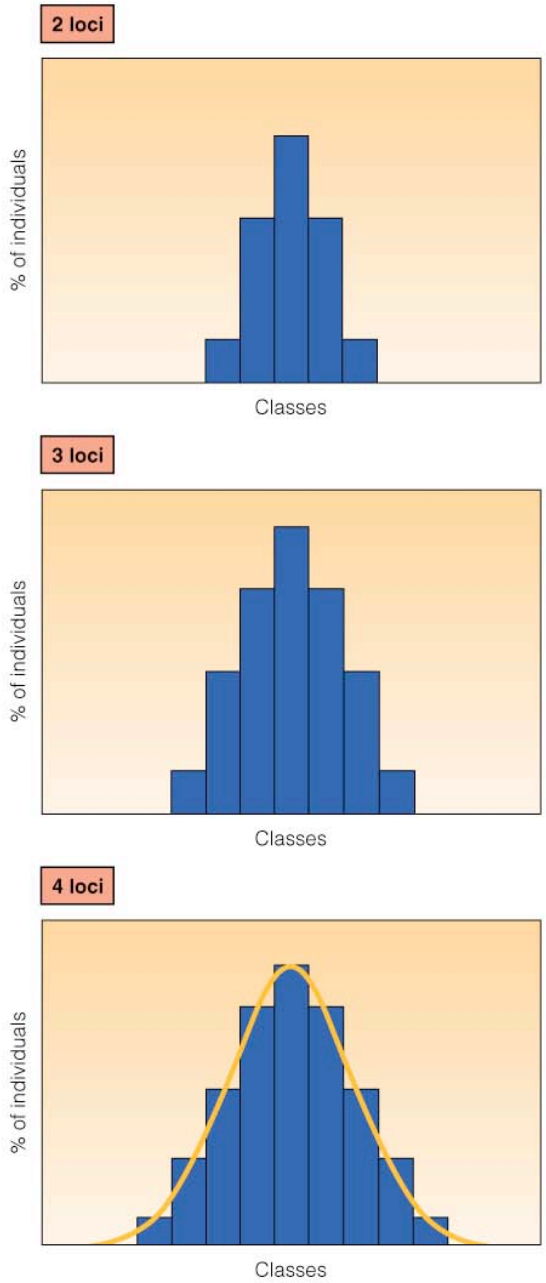
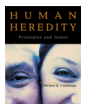
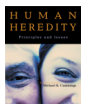


Fig. 5.5



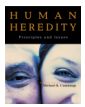
Multifactorial Traits

- Genotype does not change after fertilization (except by mutation)
- Phenotype is the sum of the observable characteristics and may change throughout life
- Environment includes all genetic and nongenetic factors



Characteristics of Multifactorial Traits

- Polygenic
- Genes controlling trait act additively
- Environmental factors interact with the genotype to produce the phenotype
- Assessing interactions can be difficult



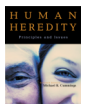
Methods Used to Study Multifactorial Traits

- **Threshold model**

  Frequency of disorder among relatives is compared with the frequency of the disorder in the general population

- **Recurrence risk**

  Estimates the risk that the disease will recur



Threshold Model

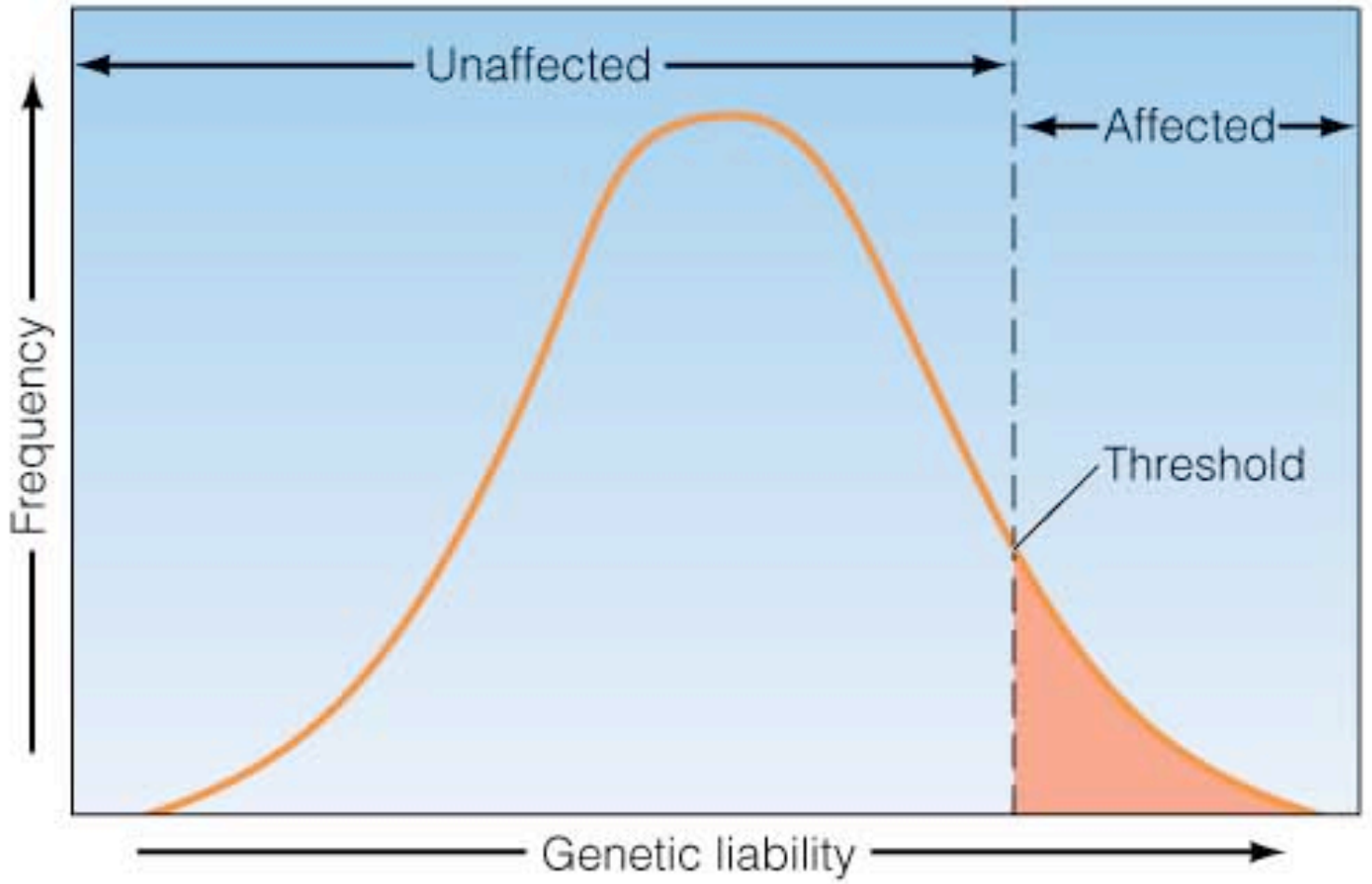
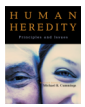


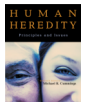
Fig. 5.7



Familial Risk

Table 5.1 Familial Risks for Multifactorial Threshold Traits

Multifactorial Trait	Risk Relative to General Population			
	MZ Twins	First-Degree Relatives	Second-Degree Relatives	Third-Degree Relatives
Clubfoot	300×	25×	5×	2.0×
Cleft lip	400×	40×	7×	3.0×
Congenital hip dislocation (females only)	200×	25×	3×	2.0×
Congenital pyloric stenosis (males only)	80×	10×	5×	1.5×

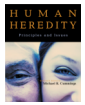


Phenotypic Variation

Sources of phenotypic variation

- Genotypes in the population
- Variation in the environment

Heritability – how much of the observed phenotypic variation is due to differences in genotype



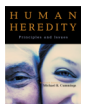
Factors that Contribute to Phenotypic Variance

- **Genetic variance**

  Variance attributed to the genotypic differences
- **Environmental variance**

  Variance attributed to differences in the environment
- **Correlation coefficients**

  Measure the degree to which variables vary together



Heritability of Fingerprints

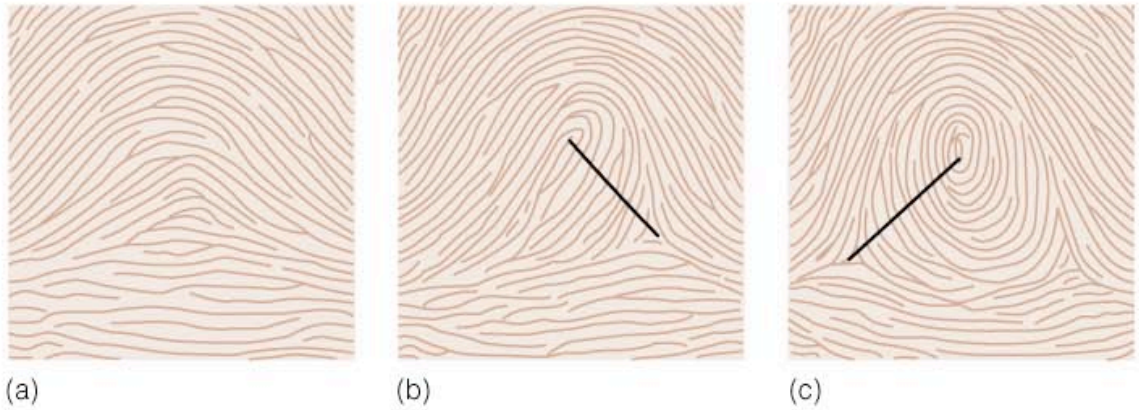
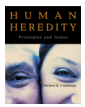


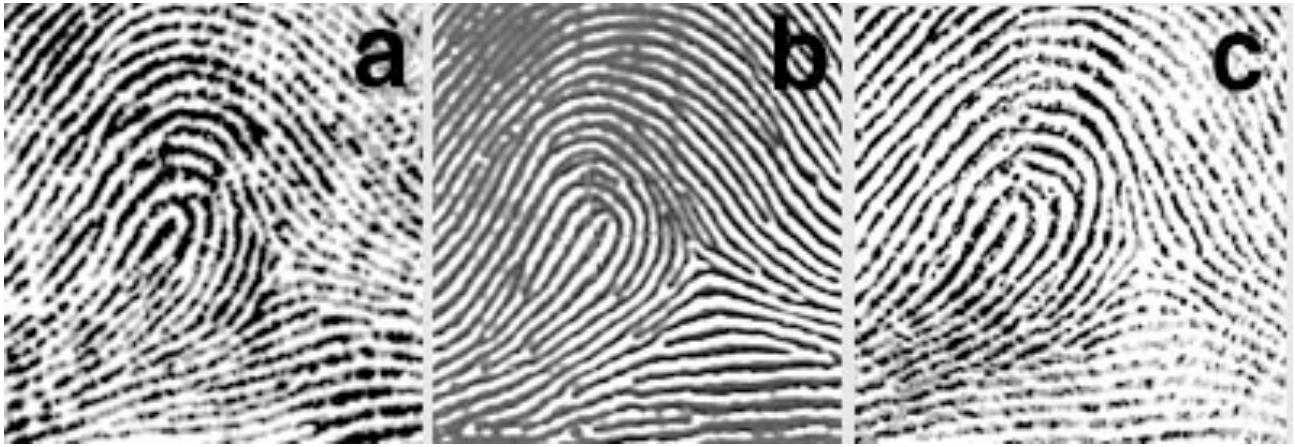
Fig. 5.8

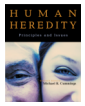
Table 5.2 Correlations between Relatives for Total Ridge Count (TRC)

Relationship	Number of Pairs	Observed Correlation Coefficient	Expected Correlation Coefficient between Relatives	Heritability
Mother-child	405	0.48 ± 0.04	0.50	0.96
Father-child	405	0.49 ± 0.04	0.50	0.98
Husband-wife	200	0.05 ± 0.07	0.00	—
Sibling-sibling	642	0.50 ± 0.04	0.50	1.00
Monozygotic twins	80	0.95 ± 0.01	1.00	0.95
Dizygotic twins	92	0.49 ± 0.08	0.50	0.98

Note: From *Quantitative genetics of fingerprint patterns*, by S. B. Holt (1961). *Br. Med. Bull.*, 17, 247–250.




a
b
c



Twin Studies

- **Monozygotic twins**
  - Single fertilization
  - Genetically identical

- **Dizygotic twins**
  - Independent fertilizations
  - Share approximately half their genes

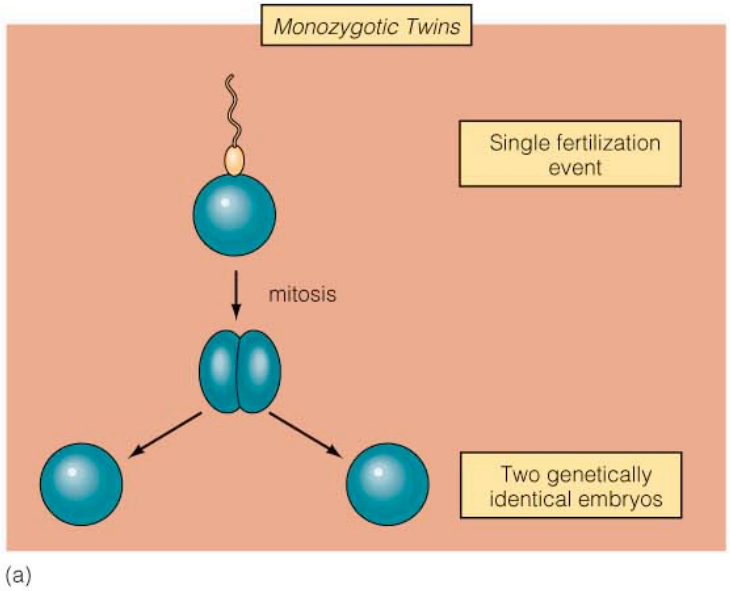


Fig. 5.10