Polygenic and Multifactoral Traits

- **Polygenic inheritance**

Continuous variation

Additive alleles

Calculating the number of genes

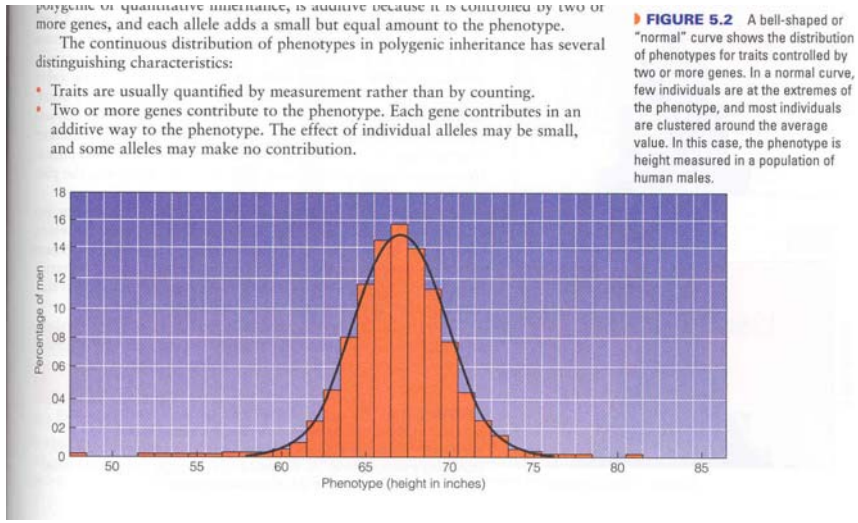
- **Heritability**

Statistical tools: Mean, variance

Broad sense heritability

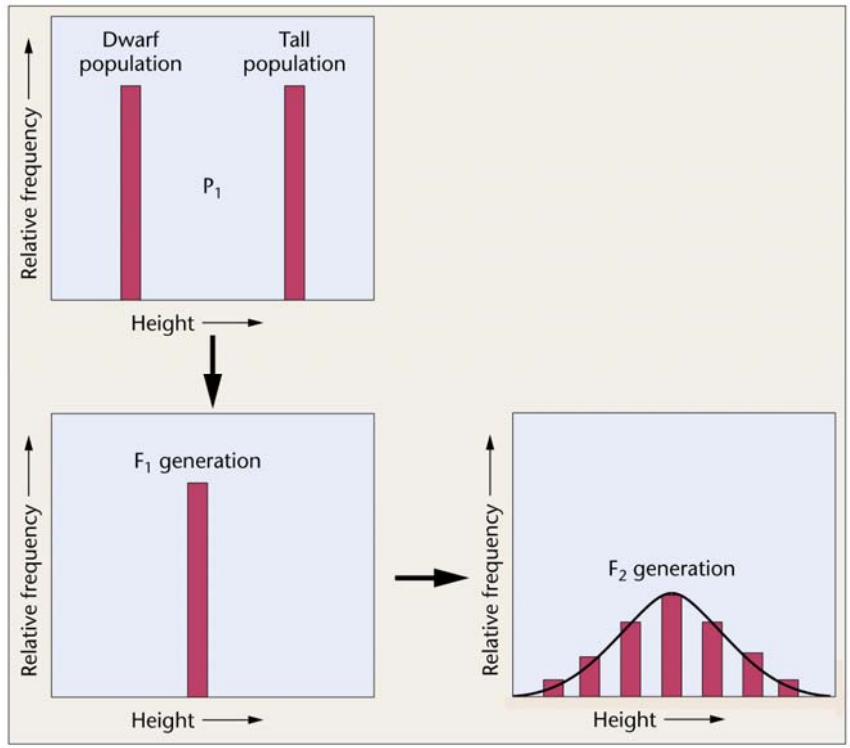
Narrow sense heritability

Twin Studies and concordance



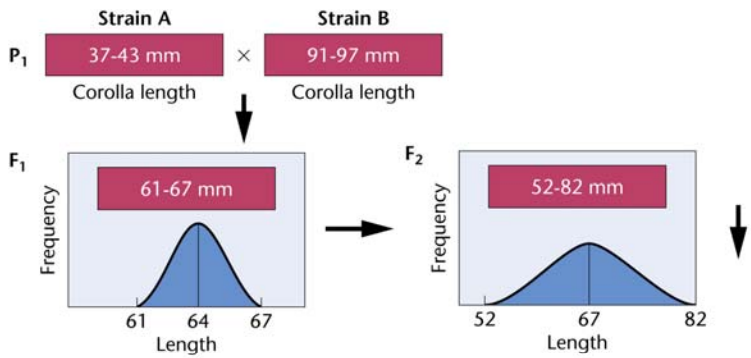
Continuous variation

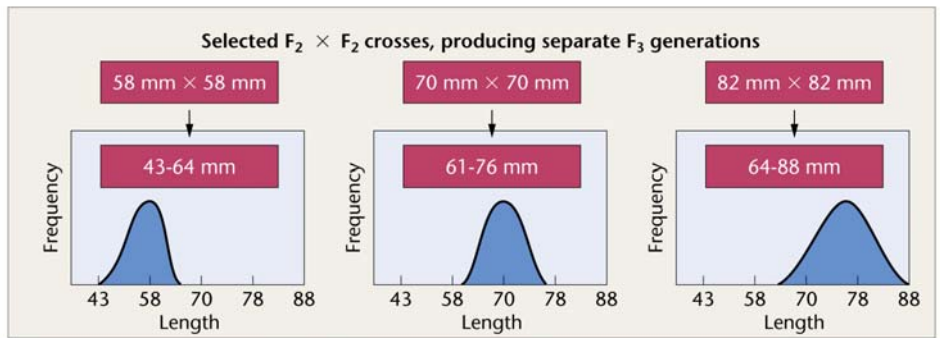
- Kolreuter's cross
- Dwarf x tall tobacco
- F1 intermediate
- F2 intermediate, normal distribution



Multiple gene hypothesis

- East's cross of Nicotiana with different corolla length
- Indicates Mendelian segregation of different phenotypic classes
- Took subsets of F_2 and crossed.



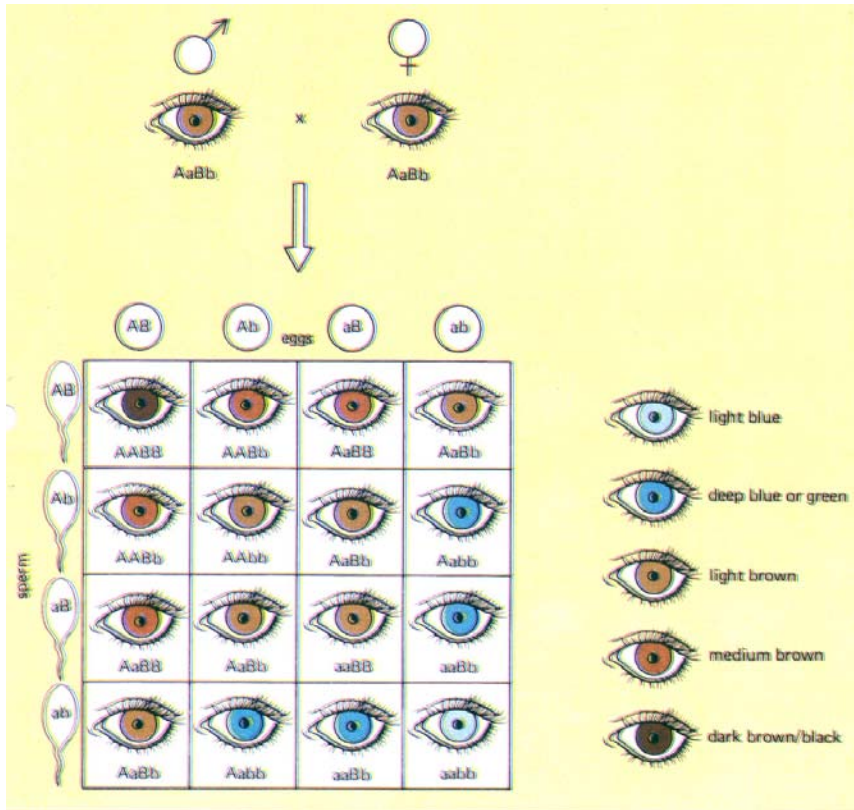


Multiple factor hypothesis

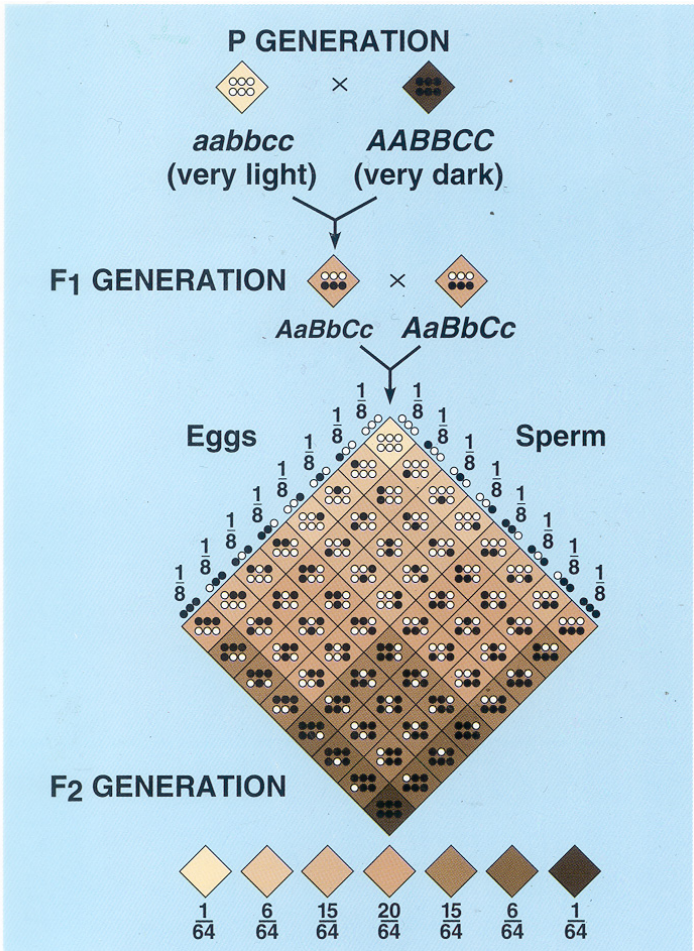
- Characters quantified
- Two or more genes
- Additive alleles
- Contribute a constant amount
- Non-additive add nothing
- All alleles add equally

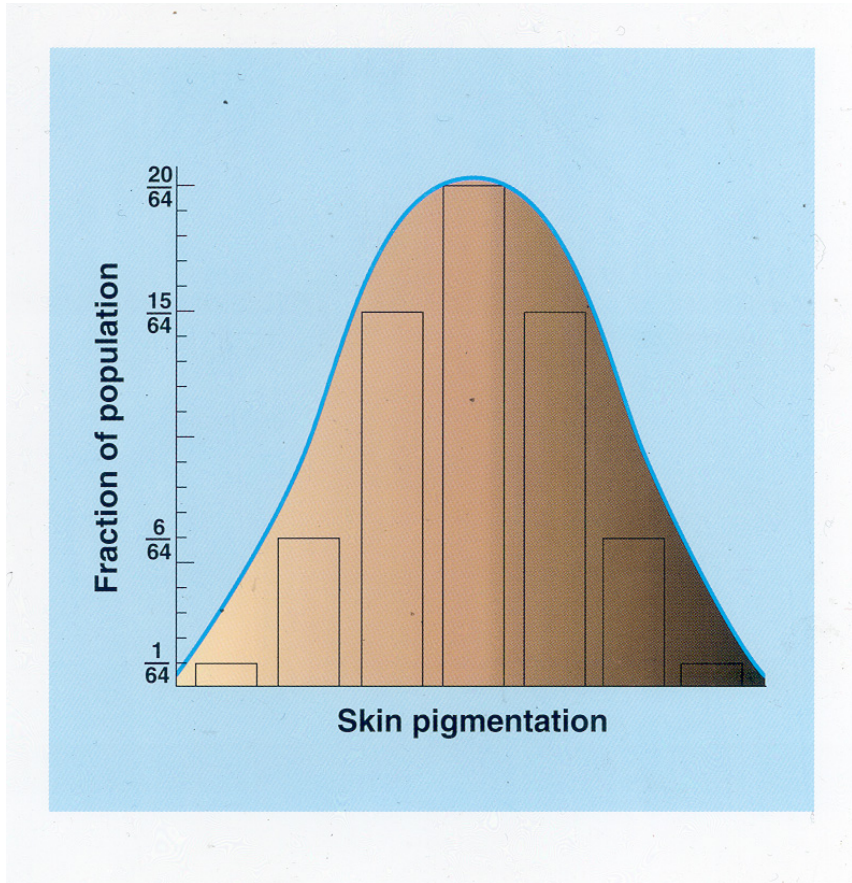
Polygenic inheritance

- 2 or more genes
- Show continuous variation vs discontinuous
- Additive component
- Distinct phenotypic classes
- Quantitative traits: size, weight, height,IQ



Polygenic inheritance





Skin color

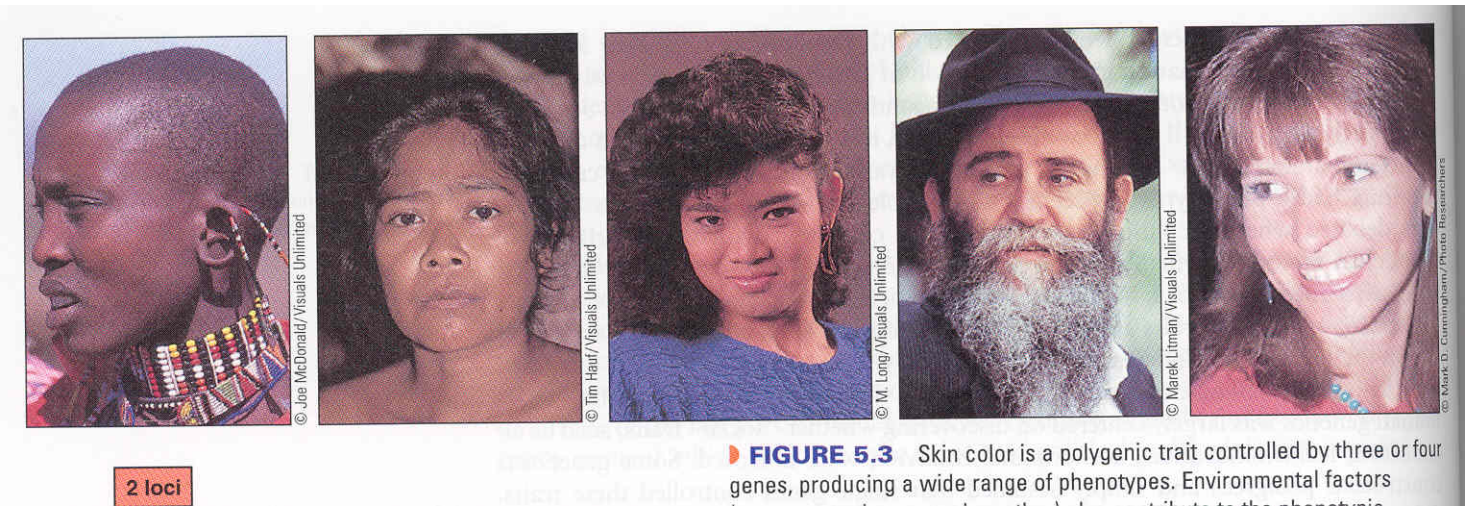
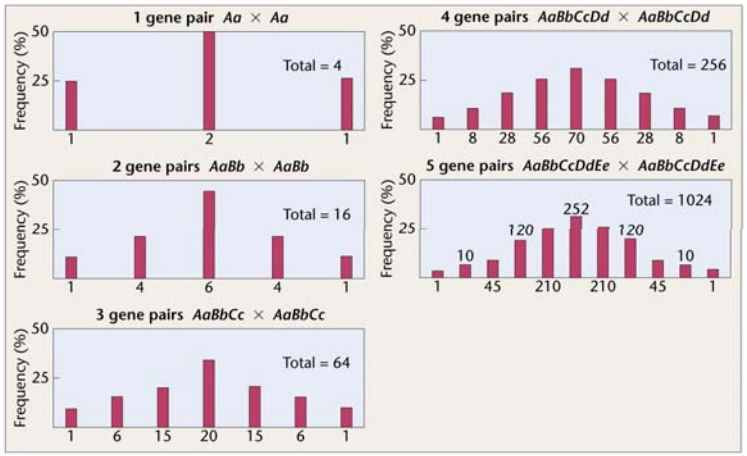


FIGURE 5.3 Skin color is a polygenic trait controlled by three or four genes, producing a wide range of phenotypes. Environmental factors

Additive Model

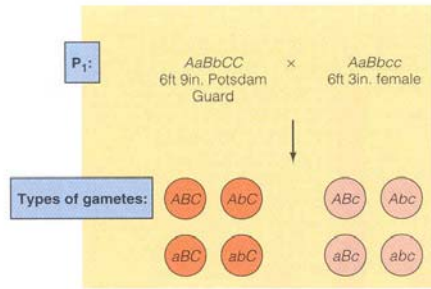


- $(1/4)^n$= ratio of f_2 individuals showing extreme phenotype
- n = (2n +1) phenotypic classes

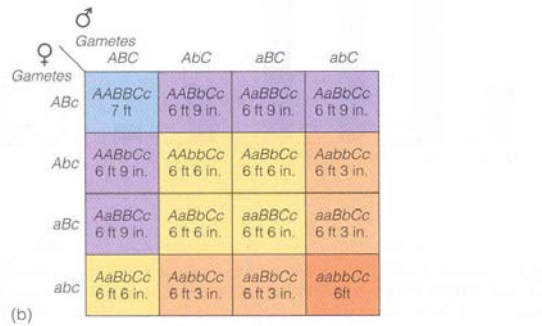
Regression to the mean

- Tendency of offspring of parents with extreme differences in phenotype to exhibit a phenotype that is average of the two parental phenotypes
- Dominant and additive effects

FIGURE 5.7 A model for inheritance of height in the Potsdam Guards. In this example, the guards and their mates represent a subset of individuals in a population in which height can range from 5 ft. 9 in. (*aabbcc*) to 7 ft. 3 in. (*AABBCC*). (a) Gametes produced by a 6 ft. 9 in. male and a 6 ft. 3 in. female. (b) Punnett square showing the 16 genotypic and 5 phenotypic combinations that result from fertilization of all combinations of gametes. The genotypes resulting in children who are as tall or taller than their father are noted. Most of the children will have a height intermediate to their parents, showing regression to a mean height.



(a)



(b)

Threshold effects

- Some individuals show affected or unaffected phenotypes
- Predisposition is caused by a number of genes in an additive way
- Will develop the genetic disorder if exposed to proper environmental conditions.

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ultifactorial Traits

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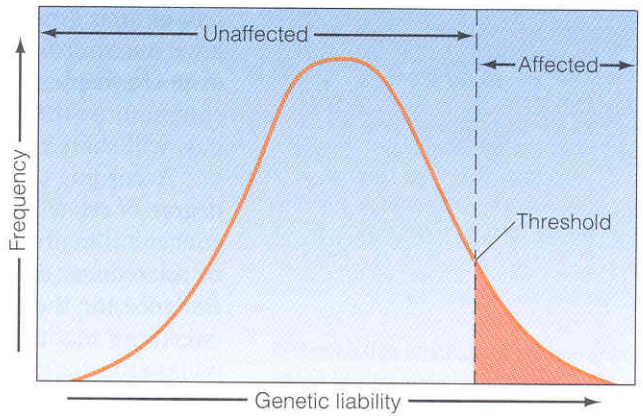
FIGURE 5.9 A model to explain the discontinuous distribution of some multifactorial traits. In this model, liability for a genetic disorder is distributed among individuals in a normal curve. This liability is caused by a number of genes, each acting additively. Only those individuals who have a genetic liability above a certain threshold are affected if exposed to certain environmental conditions.



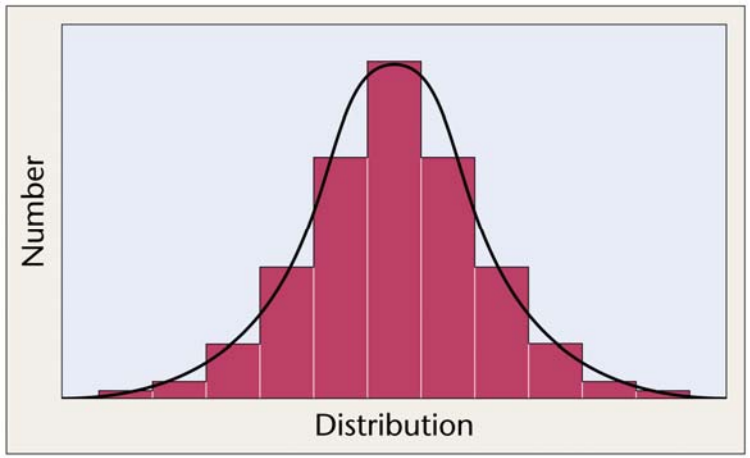
Familial Risks

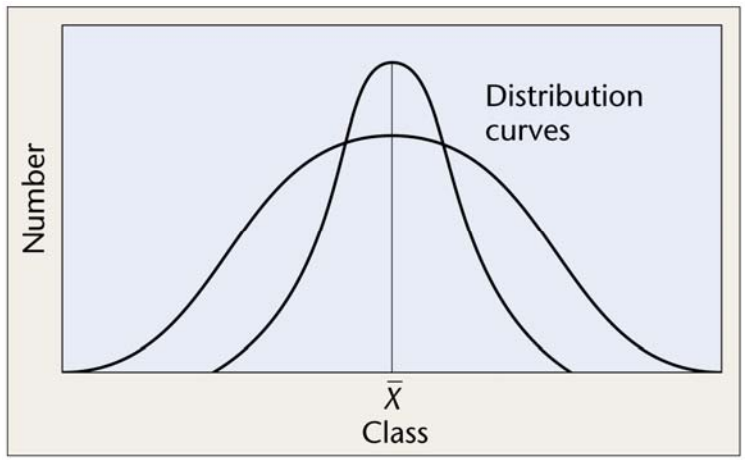
- First degree: Parent-child share ½ genes
- Second degree: grandfather: grandchild share ¼ genes
- Third degree share 1/8 genes

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
Club foot	300×	25×	5×	2×
Cleft lip	400×	40×	7×	3×
Congenital hip dislocation (females only)	200×	25×	3×	2×
Congenital pyloric stenosis (males only)	80×	10×	5×	1.5×

Statistical Analysis





- Mean (X) = $\sum X_i/n$
- Variance (s^2) = $(\sum X_i - X)^2/n\text{-}1$
- Standard deviation (s) = $\sqrt{s^2}$

Broad-sense Heritabilty

- H^2 = proportion of total variance caused by genetic variance
- H^2 = 1.0, all genetic
- H^2 = 0 all variation due to environment

- Vp= phenotypic var
- Vg= genetic var
- V_E= enviromental var
- $Vp = Vg + V_E$
- $H^2 = Vg/Vp$

Twin studies

Table 5.3 Concordance Values in Monozygotic (MZ) and Dizygotic (DZ) Twins

Trait	Concordance Values (%) MZ	DZ
Blood types	100	66
Eye color	99	28
Mental retardation	97	37
Hair color	89	22
Down syndrome	89	7
Handedness (left or right)	79	77
Epilepsy	72	15
Diabetes	65	18
Tuberculosis	56	22
Cleft lip	42	5

Table 5.4 Concordance Values for Obesity in Twins

% Overweight	% Concordant at Military Induction		% Concordant 25 Years Later	
	MZ	DZ	MZ	DZ
15	61	31	68	49
20	57	27	60	40
25	46	24	54	26
30	51	19	47	16
40	44	0	36	6

Note: From A twin study of human obesity, by A. J. Stunkard, T. T. Foch, and Z. Hrubec, (1986). JAMA, *256, 51–54.*

Heritability

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	1.0
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.*

Table 5.5 Heritability Estimates for Obesity in Twins (from Several Studies)

Condition	Heritability
Obesity in children	0.77–0.88
Obesity in adults (weight at age 45)	0.64
Obesity in adults (body mass index at age 20)	0.80
Obesity in adults (weight at induction into armed forces)	0.77
Obesity in twins reared together or apart	
Men	0.70
Women	0.66

Survey of traits

FIGURE 5.24 A graphical representation of correlations in IQ measurements in different sets of individuals. The expected correlation coefficients are determined by the degree of genetic relatedness in each set of individuals. The vertical line represents the median correlation coefficient in each case.

population. Heritability differences between two populations cannot be compa
because heritability measures only variation within a population at the time of m
surement and cannot be used to estimate genetic variation between populations
other words, we cannot use heritability differences between groups to conclude t
there are genetic differences between those groups.

It is quite evident that both genetic and environmental factors make impor
contributions to intelligence. Clearly, the relative amount each contributes canno

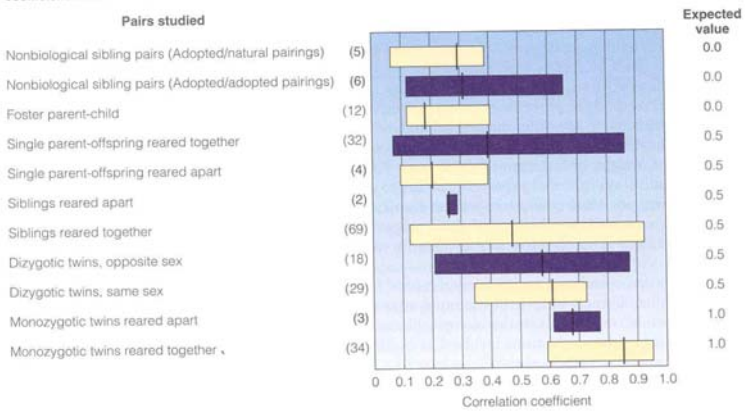


Table 9.2 Quasi-continuous multifactorial phenotypes

Group	Trait	Brief description
Congenital heart defects	Atrial septal defects	Hole in the septum of the atrium
	Coarctation of aorta	Narrowing of the aorta
	Patent ductus arteriosis	Failure of duct closure between the aorta and pulmonary arteries
	Pulmonary stenosis	Narrowing of the pulmonary artery
	Translocated great arteries	Exchange position of the two major arteries
	Ventricular septal defects	Hole in the septum separating the ventricles
Coronary heart disease	Hypercholesterolemia	High blood levels of cholesterol
	Hypertriglyceridemia	High blood levels of triglycerides
	Combined hyperlipidemia	High blood lipid levels
	Arteriosclerosis	Hardening of the coronary arteries
	Essential hypertension	High blood pressure
Diabetes	Maturity onset—noninsulin dependent	Metabolic carbohydrate disorder
	Juvenile onset—insulin dependent (HLA-, DR3-, and DR4-associated)	Metabolic carbohydrate disorder
Head, feet, mouth, eyes, and ears	Hydrocephaly (one type)	Water on the brain
	Club foot	Feet turned in at birth
	Cleft lip	Incomplete formation of the lip (see fig. 9.2)
	Cleft palate	Incomplete formation of the roof of the mouth
	Cleft lip and palate	See above
	Stuttering	Speech delay in articulating letters and words
	Strabismus	Rapid movement of the eyes
	Profound deafness (some cases)	Loss of hearing
Joints	Congenital hip	Dislocation of hip from birth
	Rheumatoid arthritis	Swelling and stiffening of joints
Mental state	Alzheimer's disease	Mental disorientation
	Manic depressive psychosis	Severe alteration of moods
	Nonspecific mental retardation	Other than single gene, chromosome or environmental agent
	Schizophrenia	Split personalities
Skin and muscle	Lupus erythematosus	Disease of the connective tissue
Spine	Anencephaly	Absence of complete brain
	Ankylosing spondylitis (HLA-B27 haplotype)	Immobility of the vertebrae
	Meningomyelocele	Open spine
	Scoliosis	Curvature of the spine
Stomach, colon, kidney	Pyloric stenosis	Narrow opening from stomach to intestine
	Crohn's disease	Irritation of the ileum
	Peptic ulcer	Ulceration of a mucous membrane
	Hirschsprung's disease (megacolon)	Absence of innervation of distal colon and rectum
	Familial idiopathic nephrotic syndrome	Massive swelling due to kidney disease