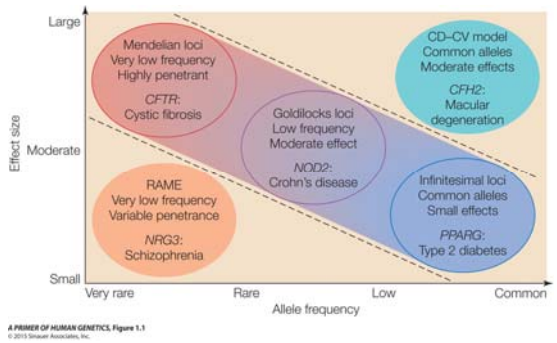
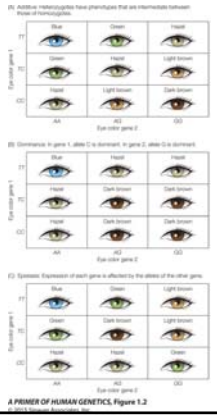
Figure 1.1 The Spectrum of Genetic Contributions to Disease



A PRIMER OF HUMAN GENETICS, Figure 1.1
© 2015 Sinauer Associates, Inc.

Figure 1.2 Additivity, Dominance, and Epistasis



A PRIMER OF HUMAN GENETICS, Figure 1.2

Figure 1.2 Additivity, Dominance, and Epistasis

Dihybrid Cross, two loci

Locus 1: C & T Locus 2: G & A

P **CCGG** x ***TTAA***

F1 **CTGA** x **CTGA**

F2	**CG**	**CA**	**TG**	**TA**
CG	CCGG	CCGA	CTGG	CTGA
CA	CCGA	CCAA	CTGA	CTAA
TG	CTGG	CTGA	*TTGG*	*TTGA*
TA	CTGA	CTAA	*TTGA*	*TTAA*

Figure 1.2 Additivity, Dominance, and Epistasis (Part 1)

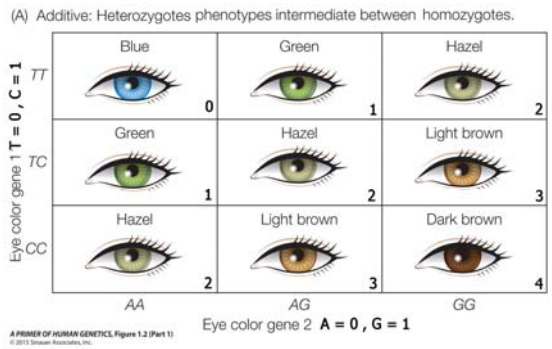


Figure 1.2 Additivity, Dominance, and Epistasis (Part 1)

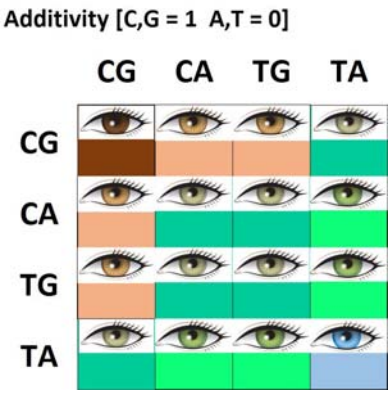


Figure 1.2 Additivity, Dominance, and Epistasis (Part 2)

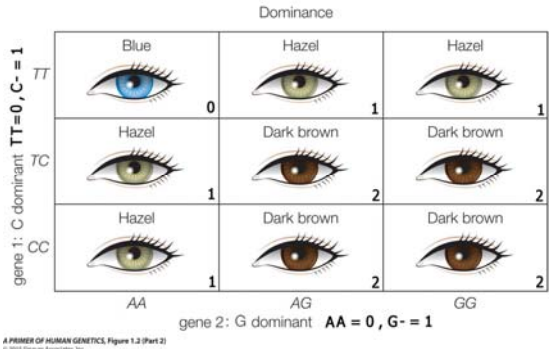


Figure 1.2 Additivity, Dominance, and Epistasis (Part 2)

Dominance (two loci) [G > A ; C > T)

	CG	CA	TG	TA
CG	+ +	+ +	+ +	+ +
CA	+ +	- +	+ +	- +
TG	+ +	+ +	+ -	+ -
TA	+ +	- +	+ -	- -

Figure 1.2 Additivity, Dominance, and Epistasis (Part 3)

(C) Epistasis

Eye color gene 1 \ Eye color gene 2	AA	AG	GG
TT	Blue AA & TT	Green not GG & not CC	Light brown GG
TC	Green not GG & not CC	Dark brown AG & CT	Light brown GG
CC	Hazel CC	Hazel CC	Light brown GG >CC

A PRIMER OF HUMAN GENETICS, Figure 1.2 (Part 3)
© 2015 Sinauer Associates, Inc.

Figure 1.2 Additivity, Dominance, and Epistasis (Part 3)

Epistasis [locus x locus interaction]

	CG	CA	TG	TA
CG	GG >CC	CC	GG	AG&CT
CA	CC	CC	AG&CT	not GG not CC
TG	GG	AG&CT	GG-	not GG not CC
TA	AG&CT	not GG not CC	not GG not CC	AA&TT

Figure 1.3 Estimation of Heritability

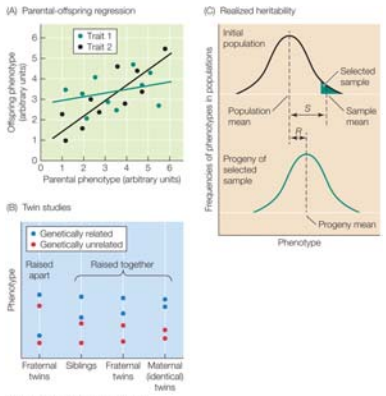
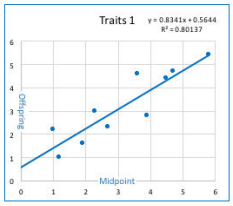


Figure 1.3 Estimation of Heritability (Part 1)

	Midpoint	Offspring
1	1.0	2.2
2	1.2	1.0
3	1.9	1.6
4	2.3	3.0
5	2.7	2.3
6	3.6	4.6
7	3.9	2.8
8	4.5	4.4
9	4.7	4.7
10	5.8	5.4



	Midpoint	Offspring
1	1.2	3.4
2	1.9	3.2
3	2.1	2.0
4	2.6	4.1
5	3.1	2.9
6	3.5	2.5
7	4.3	4.7
8	4.7	3.7
9	5.1	4.3
10	5.5	2.7

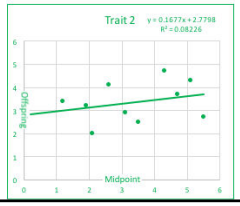


Figure 1.3 Estimation of Heritability (Part 1)

	Mid-parent	Mid-offspring
	10.26	11.12
	10.45	10.64
	10.61	10.70
	10.92	10.73
	10.95	10.66
	11.07	10.80
	11.07	10.91
	10.96	10.94
	10.97	11.03
	11.27	11.23
	11.37	11.40
	11.58	10.92
	11.79	11.66
mean	11.02	10.98
st dev	0.428	0.306

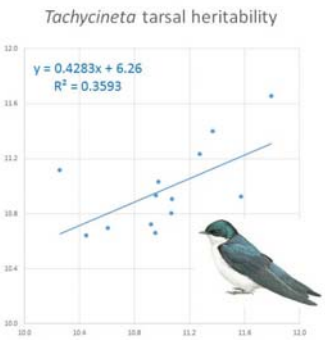


Figure 1.3 Estimation of Heritability (Part 1)

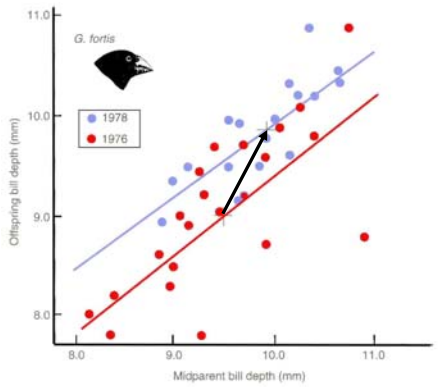


Figure 1.3 Estimation of Heritability (Part 2)

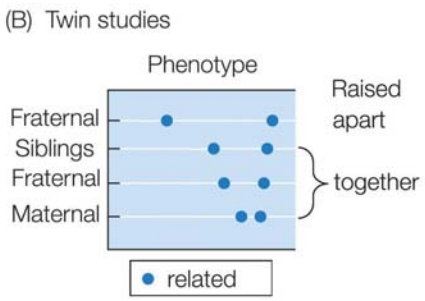
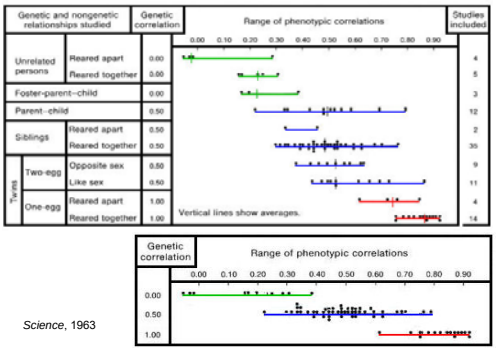


Figure 1.3 Estimation of Heritability (Part 2)



Science, 1963

Figure 1.3 Estimation of Heritability (Part 2)

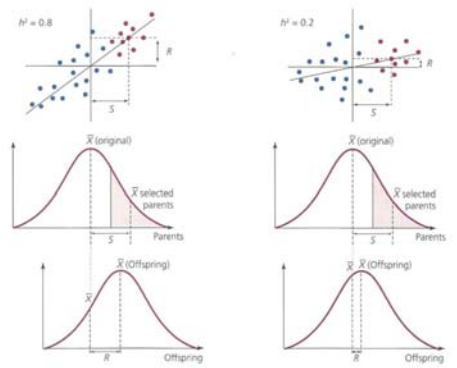
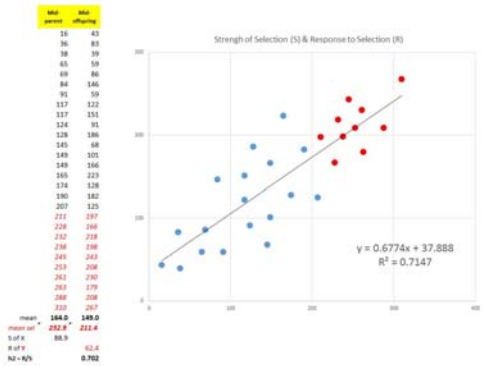
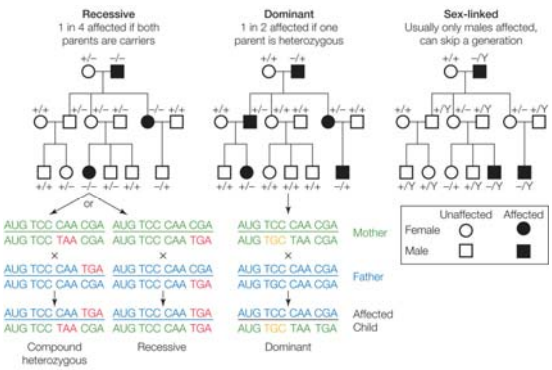


Figure 1.3 Estimation of Heritability (Part 3)



Mid-parent	Mid-offspring
16	43
36	83
38	39
65	59
69	86
84	146
91	59
117	122
117	151
124	91
128	186
145	68
149	101
149	166
165	223
174	128
190	182
207	125
211	197
228	160
232	218
236	198
245	243
253	208
261	230
263	179
288	208
310	267
mean	164.0	149.0
mean sel	252.9	211.4
S of x	88.9	
R of y		62.4
h2 = R/S		0.702

Figure 1.4 Mendelian Transmission within a Three-Generation Pedigree



A PRIMER OF HUMAN GENETICS, Figure 1.4
© 2013 Sinauer Associates, Inc.

Figure 1.4 Mendelian Transmission within a Three-Generation Pedigree

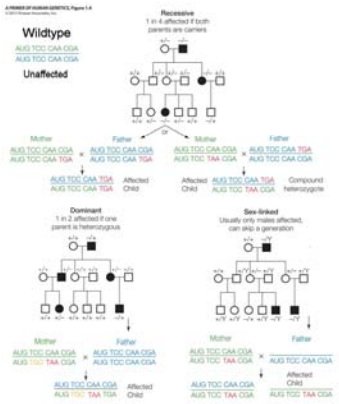
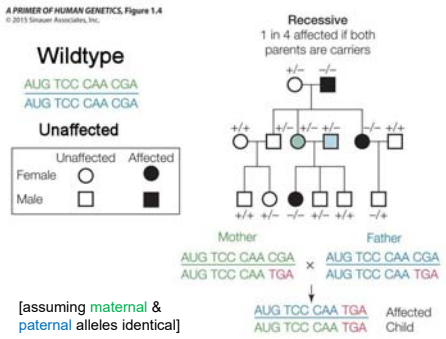
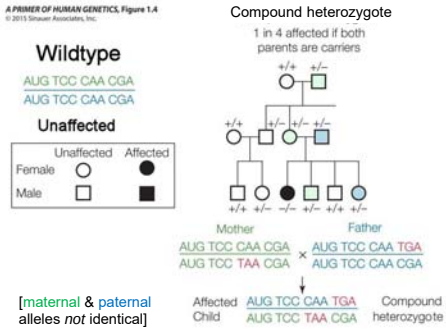


Figure 1.4 Mendelian Transmission within a Three-Generation Pedigree



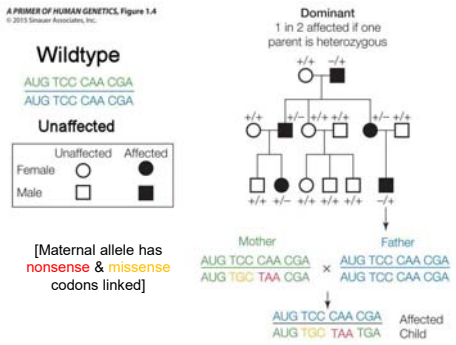
[assuming maternal & paternal alleles identical]

Figure 1.4 Mendelian Transmission within a Three-Generation Pedigree



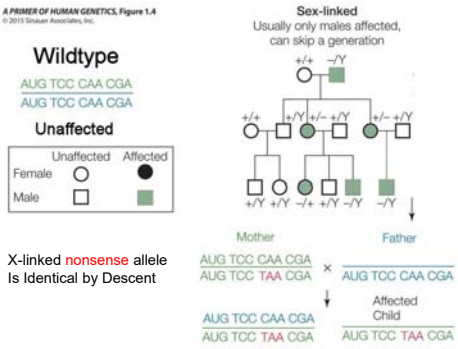
[maternal & paternal alleles *not* identical]

Figure 1.4 Mendelian Transmission within a Three-Generation Pedigree



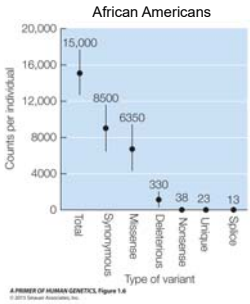
[Maternal allele has nonsense & missense codons linked]

Figure 1.4 Mendelian Transmission within a Three-Generation Pedigree



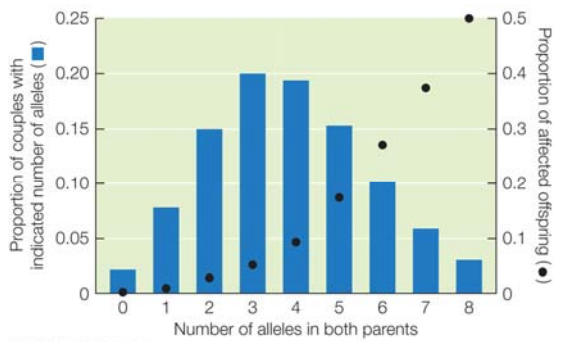
X-linked nonsense allele Is Identical by Descent

Figure 1.6 Distribution of Deleterious Mutations per Genome



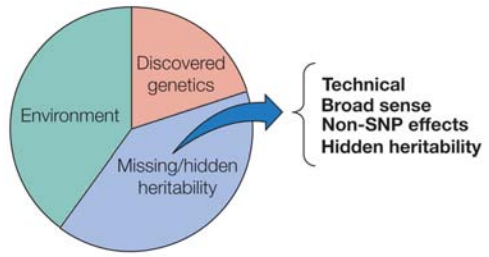
SNP Variant	African American		European American	
Synonymous	8,500	55.72%	7,000	55.47%
Mis-sense	6,350	41.63%	5,250	41.60%
Deleterious	330	2.16%	300	2.38%
Non-sense	38	0.25%	33	0.26%
Unique	23	0.15%	25	0.20%
Splice Site	13	0.09%	11	0.09%
TOTAL	15,254	100.00%	12,619	100.00%

Figure 1.7 Multiplicative Effects of Rare Mutations in Families



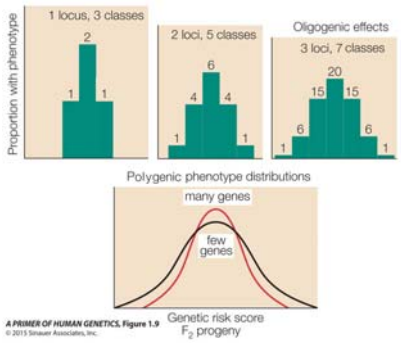
A PRIMER OF HUMAN GENETICS, Figure 1.7
© 2015 Sinauer Associates, Inc.

Figure 1.8 The Missing Heritability Problem



A PRIMER OF HUMAN GENETICS, Figure 1.8
© 2015 Sinauer Associates, Inc.

Figure 1.9 The Genetic Basis of Complex Phenotypes



A PRIMER OF HUMAN GENETICS, Figure 1.9
© 2015 Sinauer Associates, Inc.

Figure 1.10 SNP-based Heritability Estimation

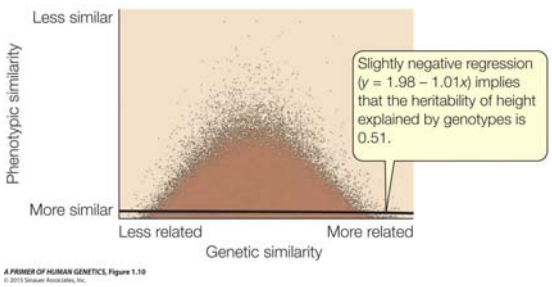


Figure 1.10 SNP-based Heritability Estimation

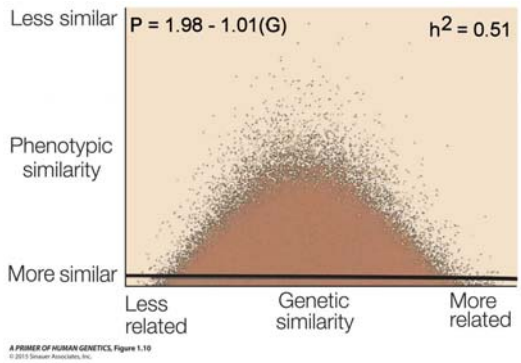
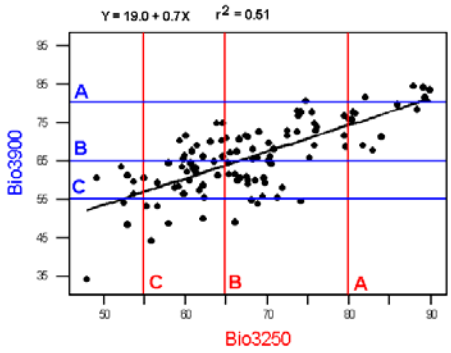



Figure 1.10 SNP-based Heritability Estimation