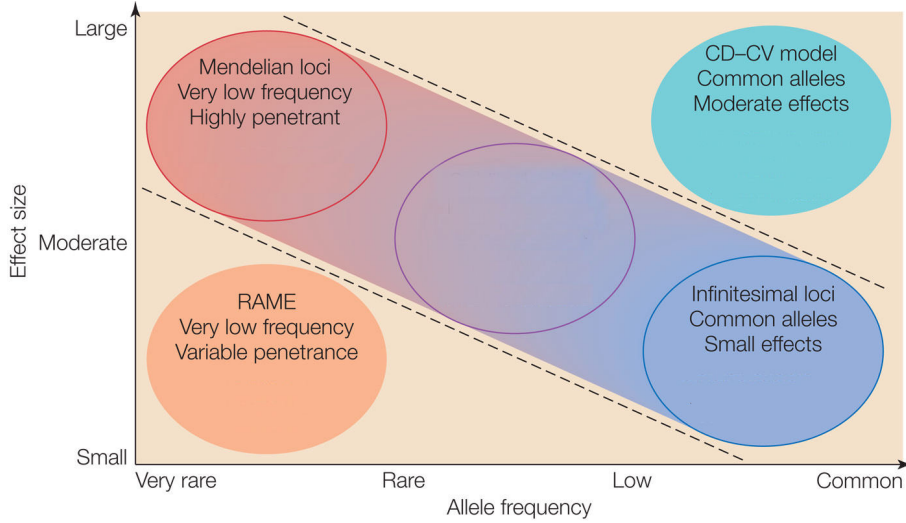


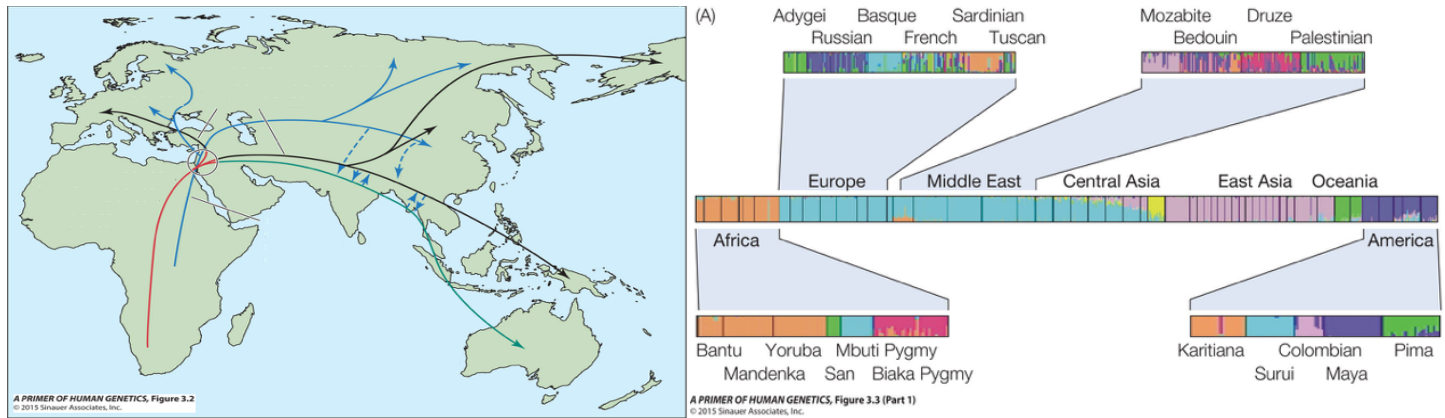
Biol4241 – Final exam supporting material

Fig 1.1 – Mendelian vs Infinitesimal Models; RAME & CD-CV



A PRIMER OF HUMAN GENETICS, Figure 1.1
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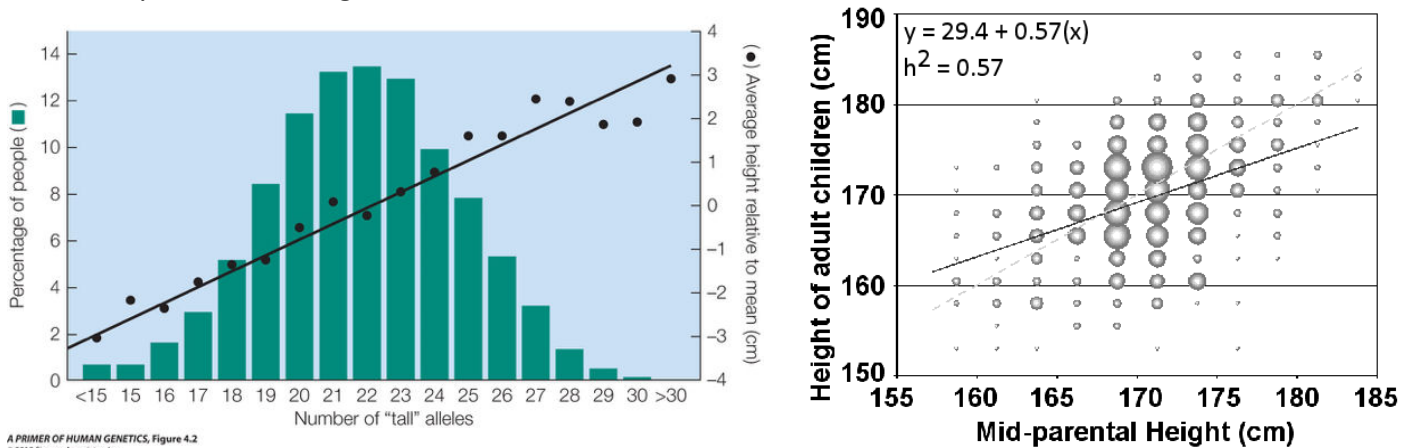
Origins of Human Continental Groups



A PRIMER OF HUMAN GENETICS, Figure 3.2
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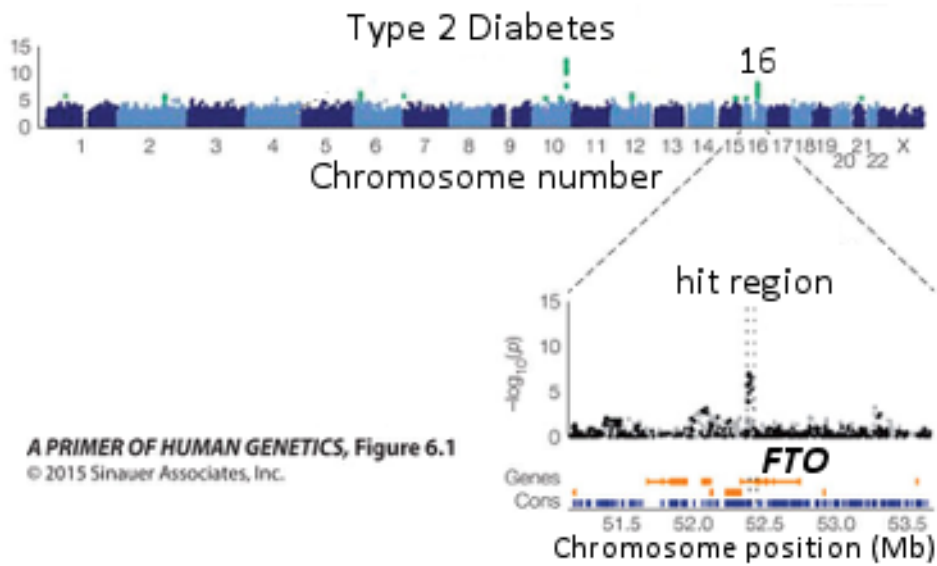
A PRIMER OF HUMAN GENETICS, Figure 3.3 (Part 1)
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Heritability of Human Height



A PRIMER OF HUMAN GENETICS, Figure 4.2
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GWAS study of Type 2 Diabetes



Patterns of Individual variation from 1,000 Genotype Project

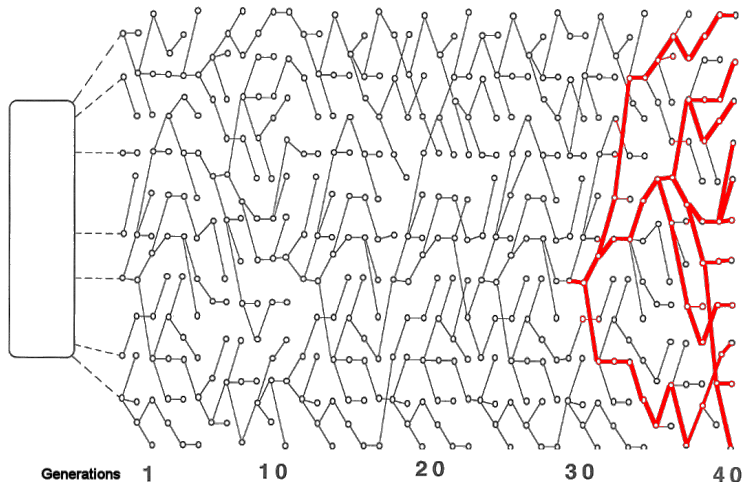
TABLE 2.1 Load of sequence variants from 1000 Genomes Project

Variant type	Number of derived sites per individual	
	<0.05% (Very rare)	>5% (Common)
All sites	30,000–150,000	3.6–3.9 million
Synonymous	29–120	1300–1400
Nonsynonymous	130–400	2300–2700
Nonsense (stop codon gain)	4–10	24–28
Indel	2–4	127–138
Splice site	3–6	5–10
Constrained UTR ^a	120–430	3500–4000
Constrained ncRNA ^a	4–17	180–200
Affecting known transcription factor binding site	23–83	750–830
Other conserved ^b	2–10	130,000–150,000

Source: WTCCC 2012.

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Coalescent Model



Interaction of Heritability & Environment

Variation can be quantified

mean ± standard deviation: $\bar{x} \pm \sigma$

variance: σ^2

Variation has two sources: **genetic** (σ_G^2) & **environmental** (σ_E^2) variance

phenotypic variance $\equiv \sigma_P^2 = \sigma_G^2 + \sigma_E^2 + \sigma_{G \times E}^2$

additive variance $\equiv \sigma_A^2 = \sigma_G^2 + \sigma_E^2$

heritability $\equiv h^2 = \sigma_G^2 / \sigma_A^2 = \sigma_G^2 / (\sigma_G^2 + \sigma_E^2)$

"Heritability in the narrow sense" ignores **interaction variance** ($\sigma_{G \times E}^2$)
Identical genotypes produce different phenotypes in different environments.

Ex.: Hypothetical IQ scores change according to adoptive environment

	Biological Parents	Children	Adoptive Parents	C - B	A - C	C x B	A x C	
1	93	113	119	20	6	10509	13447	
2	96	116	120	20	4	11136	13920	
3	99	119	111	20	-8	11781	13209	
4	98	118	112	20	-6	11564	13216	
5	90	110	114	20	4	9900	12540	
6	97	117	115	20	-2	11349	13455	
7	94	114	110	20	-4	10716	12540	
8	91	111	113	20	2	10101	12543	
9	100	120	116	20	-4	12000	13920	
10	92	112	118	20	6	10304	13216	
11	95	115	117	20	2	10925	13455	
mean	95.0	115.0	115.0	20	0	10935	13224	mean
s.d.	3.32	3.32	3.32			10925	13225	xbar*ybar
						10.000	-1.273	covariance
						1.000	-0.127	r