From genotype to phenotype

Steven Salzberg

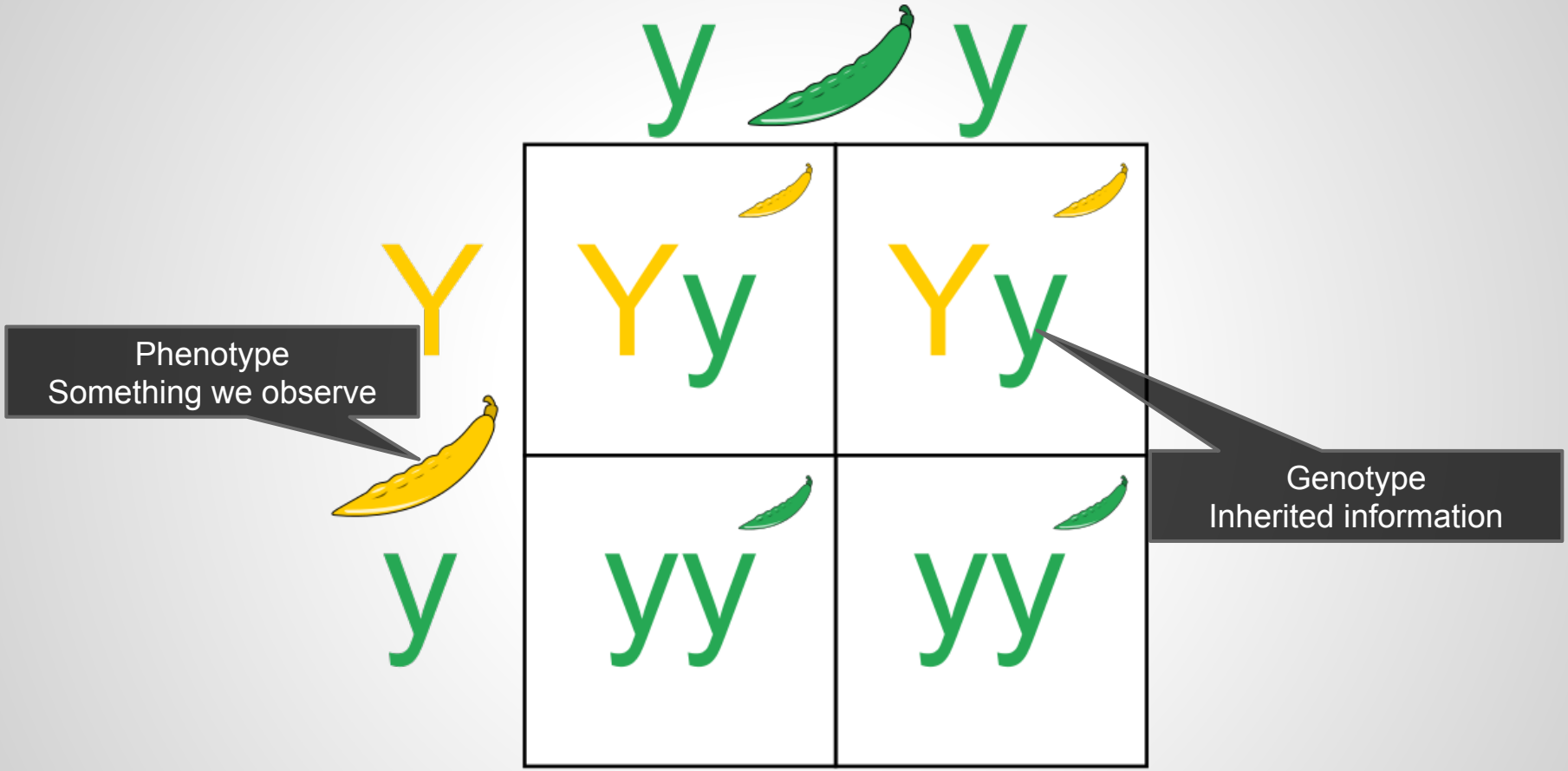


image credit: http://commons.wikimedia.org/wiki/File:Punnett_Square.svg

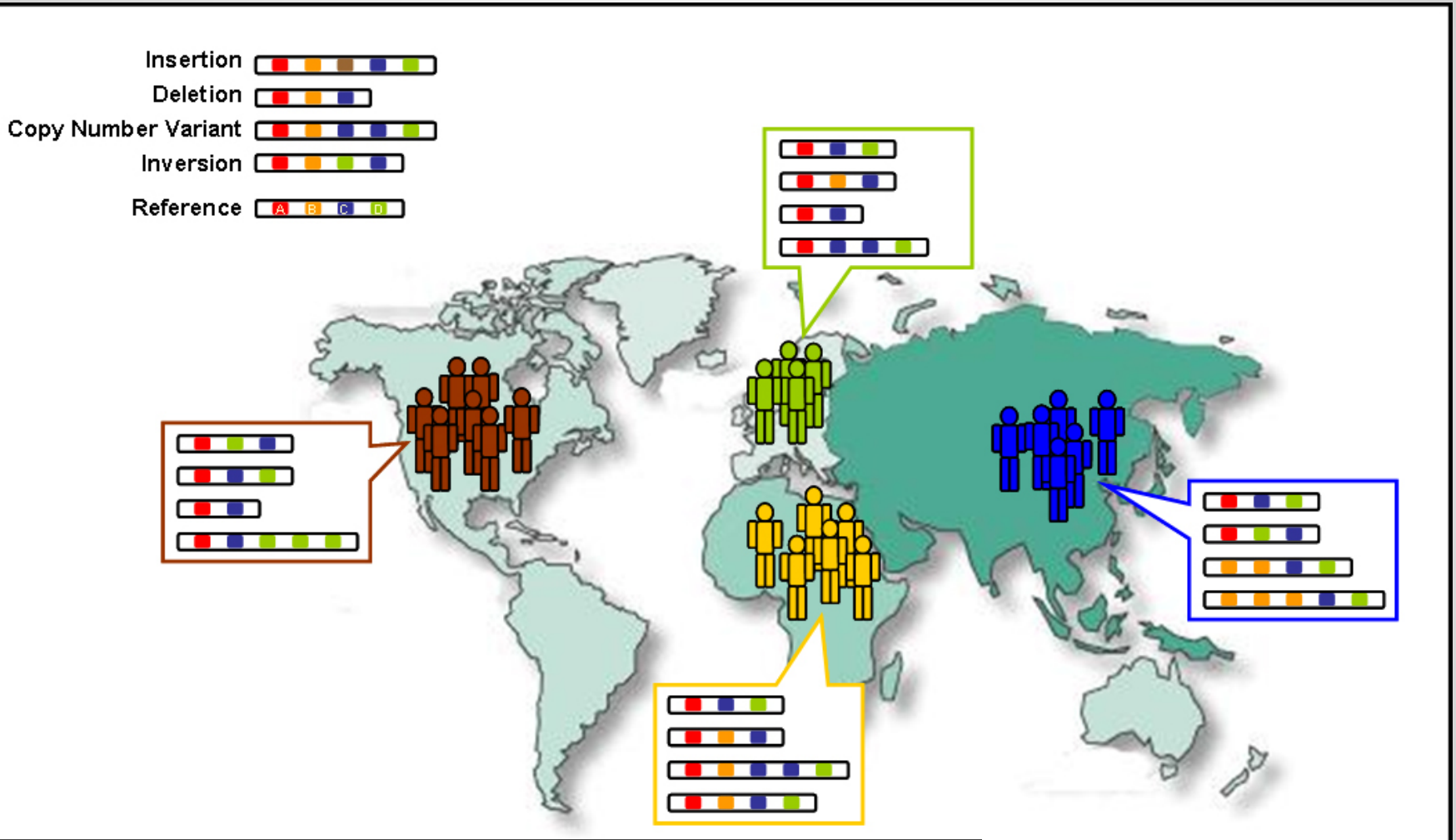


image credit: http://en.wikipedia.org/wiki/File:Genetic_Variation.jpg

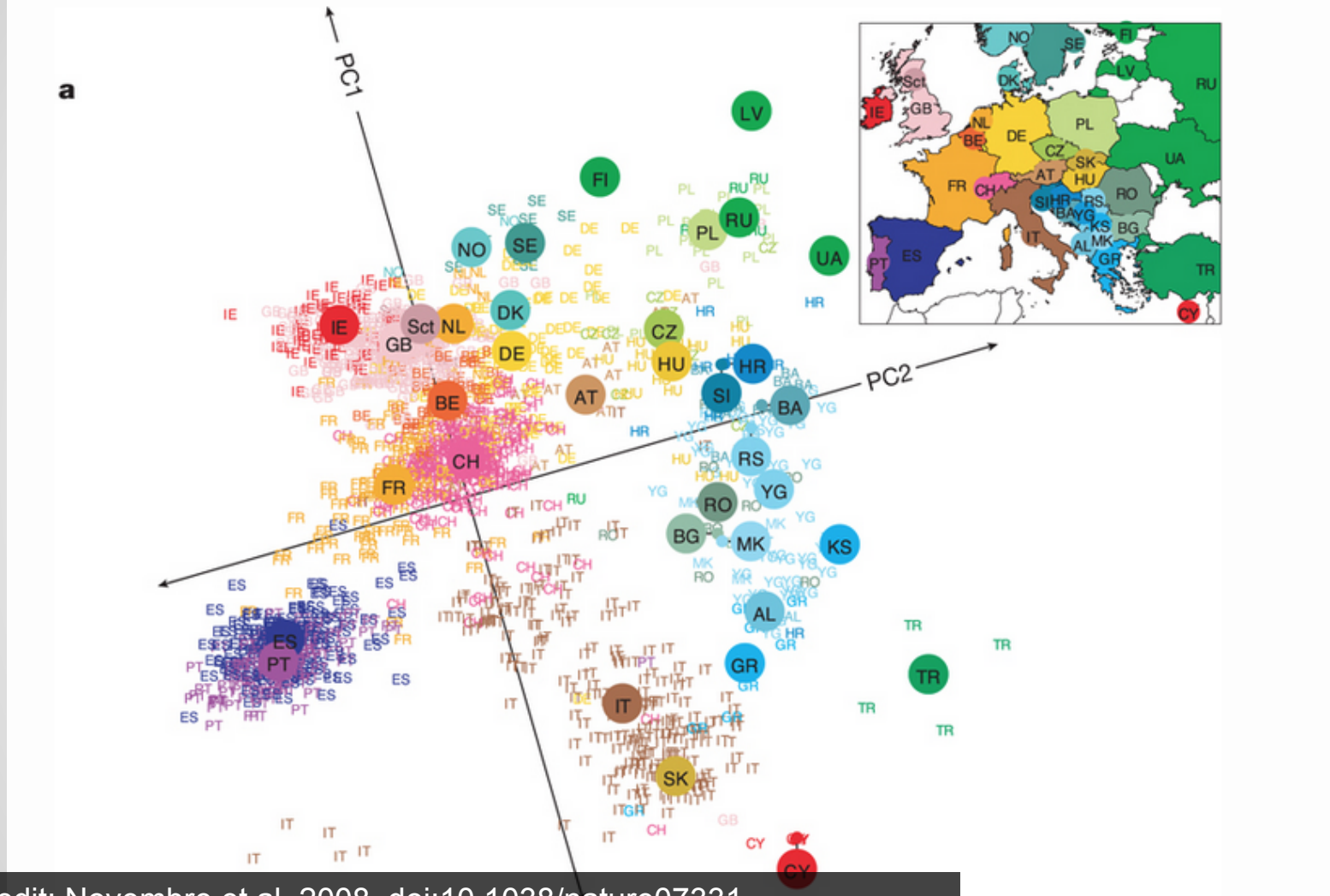
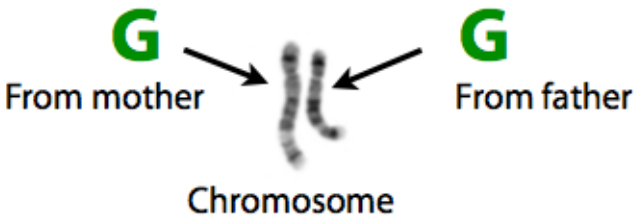


image credit: Novembre et al. 2008, doi:10.1038/nature07331

Who	Genotype	What It Means
	AA	In Europeans, 85% chance of brown eyes; 14% chance of green eyes; 1% chance of blue eyes.
	AG	In Europeans, 56% chance of brown eyes; 37% chance of green eyes; 7% chance of blue eyes.
Benjamin Langmead	GG	In Europeans, 72% chance of blue eyes; 27% chance of green eyes; 1% chance of brown eyes.

Sources: 23andme.com, Ben's genome



A and G are *alleles*

The variable site is in a gene called HERC2

Slide courtesy: Ben Langmead

Human Genetics
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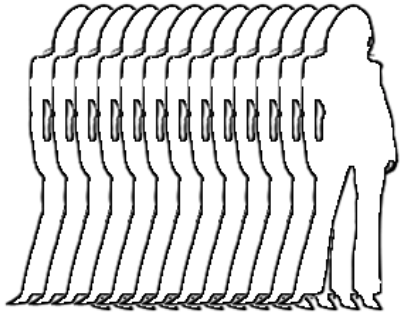
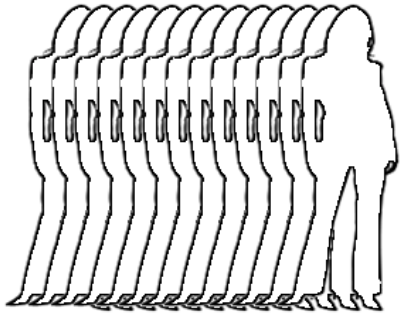
Blue eye color in humans may be caused by a perfectly associated founder mutation in a regulatory element located within the *HERC2* gene inhibiting *OCA2* expression

Hans Eiberg, Jesper Troelsen, Mette Nielsen, Annemette Mikkelsen, Jonas Mengel-From, Klaus W. Kjaer, Lars Hansen

Reference: http://link.springer.com/article/10.1007%2Fs00439-007-0460-x
Slide courtesy: Ben Langmead



GC CC GG GC CC GC GC
GG CC GC GG GC GG



GC CC GC GC GG CC CC
CC GC GC GG GC GG

SNP1	*SNP2*	*SNP …*
Cases Count of G: 2104 of 4000 Frequency of G: 52.6%	**Cases** Count of G: 1648 of 4000 Frequency of G: 41.2%	*Repeat for all SNPs*
Controls Count of G: 2676 of 6000 Frequency of G: 44.6%	**Controls** Count of G: 2532 of 6000 Frequency of G: 42.2%	
P-value: $5.0 \cdot 10^{-15}$	**P-value:** 0.33	

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