



Past and current questions? - hand in

Where we are so far....

Versions of genes (alleles) are generally handed down unaltered from generation to generation

The organisms we are primarily concerned about (humans and such) reproduce sexually, involving the processes of meiosis and fertilization

A new organism receives one set of chromosomes from each of its two parents (maternal / paternal)

It passes on a unique shuffled set (generated through crossing over and independent segregation) to each of its offspring

Where we are so far....

During meiosis, the probability of crossing over is a function of distance between two genes (in terms of base pairs)

Different versions of a gene (alleles) can lead to different phenotypes (Mendel had distinct differences, more often we have wild type and different - often disease).

Mendelian traits are strictly dominant or recessive with respect to one another.

When the alleles are of the same gene, we can generate monohybrid crosses between homozygous individuals

Q: What does it mean for a trait to "breed true"?

Where we are so far....

If two different traits are under study, our first tasks are to i) determine whether the alleles are in different genes, ii) whether the genes are linked, and iii) whether the alleles interact.

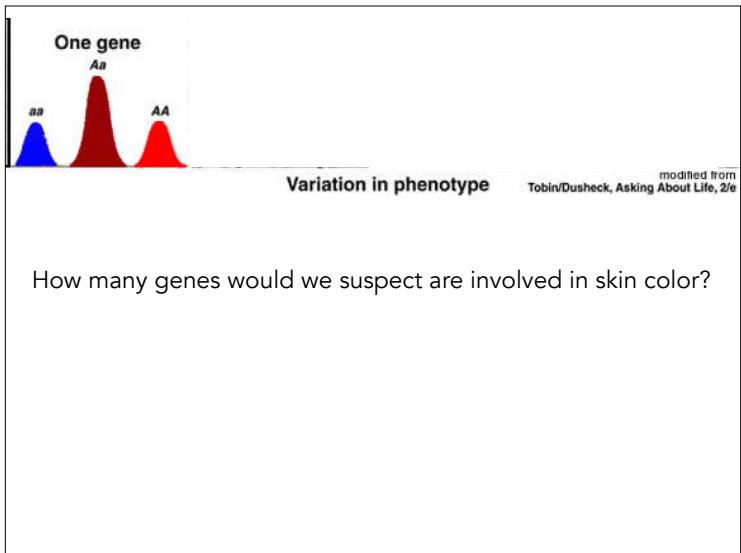
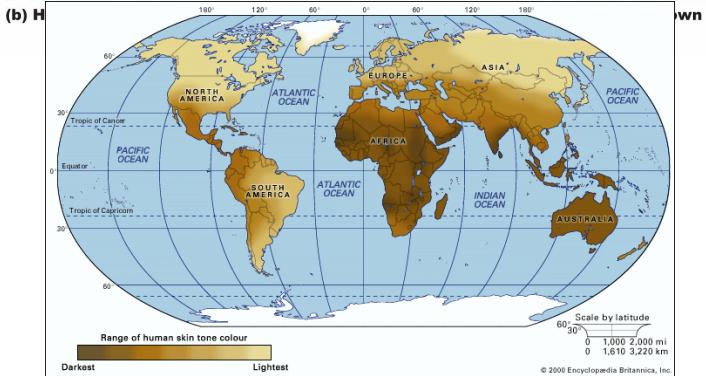
For two genes (two distinct pairs of traits), this involves a dihybrid cross. If the traits are independent, and the alleles are unlinked and strict recessive / dominant - they we get the Mendelian F₂ 9:3:3:1 ratio when we cross two (F₁) heterozygotes. (what classes are these)

If the ratio diverges from 9:3:3:1 then there is either linkage or interactions (we estimate whether the divergence is real using a chi square test).

If the ratio diverges from 9:3:3:1 then there is either linkage or interactions (we estimate whether the divergence is real using a chi square test).

What happens if they are linked?

What happens if they interact?



New gene variants reveal the evolution of human skin color

By Ann Gibbons Oct. 12, 2017, 2:00 PM



Researchers have identified genes that help create diverse skin tones, such as those seen in the Agaw (left) and Surma (right) peoples of Africa.

Researchers agree that our early australopithecine ancestors in Africa probably had light skin beneath hairy pelts. "If you shave a chimpanzee, its skin is light," says evolutionary geneticist Sarah Tishkoff of the University of Pennsylvania, the lead author of the new study. "If you have body hair, you don't need dark skin to protect you from ultraviolet [UV] radiation."

Science

RESEARCH ARTICLES

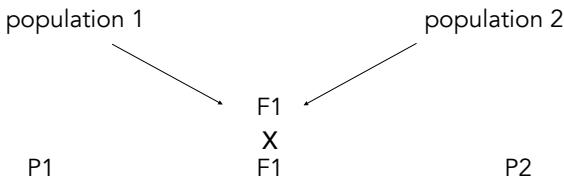
Loci associated with skin pigmentation identified in African populations

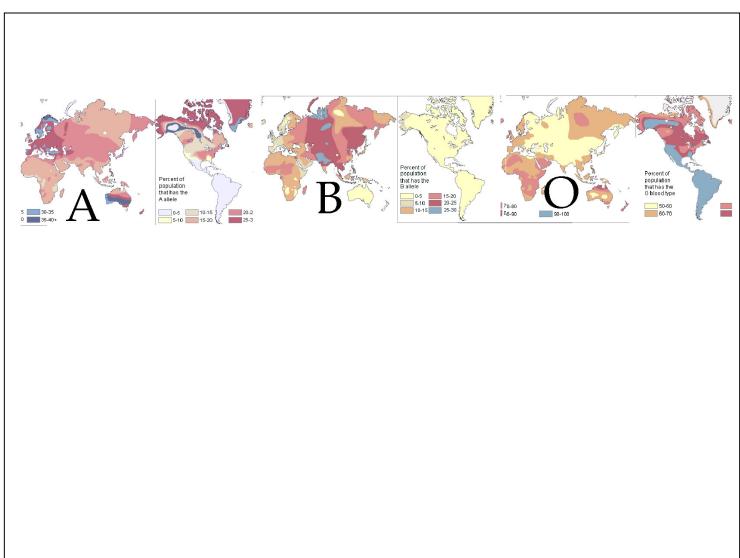
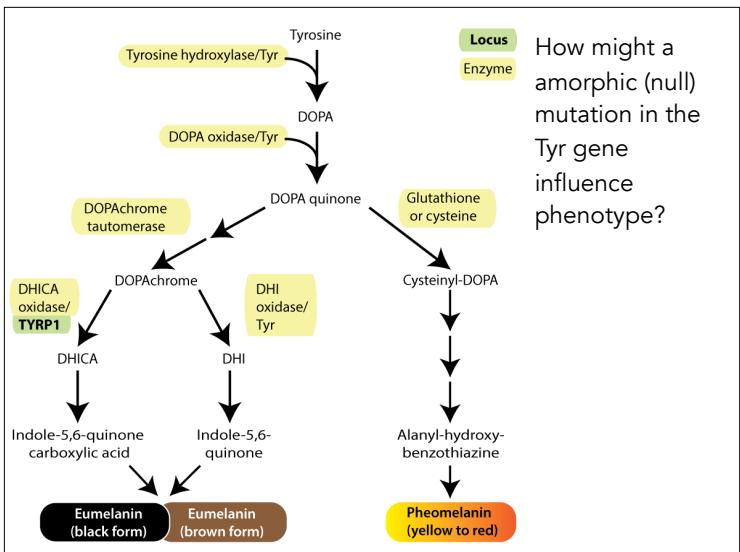
Cite as: N. G. Crawford *et al.*, *Science* 10.1126/science.aan8433 (2017).

Nicholas G. Crawford,¹ Derek E. Kelly,^{1,2*} Matthew E. B. Hansen,^{1*} Marcia H. Beltrame,^{1*} Shaohua Fan,^{1*} Shanna L. Bowman,^{3,4*} Ethan Jewett,^{5,6*} Alessia Ranciaro,¹ Simon Thompson,¹ Yancy Lo,¹ Susanne P. Pfeifer,⁷ Jeffrey D. Jensen,⁷ Michael C. Campbell,^{1,8} William Beggs,⁹ Farhad Hormozdiari,^{9,10} Sununguko Wata Mpoloka,¹¹ Gaonyadiwe George Mokone,¹² Thomas Nyambo,¹³ Davit Wold Meskel,¹⁴ Gurja Belay,¹⁴ Jake Haut,¹ NISC Comparative Sequencing Program,¹⁵ Harriet Rothschild,¹⁵ Leonard Zon,^{15,16} Yi Zhou,^{15,17} Michael A. Kovacs,¹⁸ Mai Xu,¹⁹ Tongwu Zhang,¹⁹ Kevin Bishop,¹⁹ Jason Sinclair,¹⁹ Cecilia Rivas,²⁰ Eugene Elliot,²⁰ Jiyeon Choi,¹⁸ Shengchao A. Li,^{21,22} Belynda Hicks,^{21,22} Shawn Burgess,¹⁹ Christian Abnet,²¹ Dawn E. Watkins-Chow,²⁰ Elena Oceana,²³ Yun S. Song,^{5,6,24,25,26} Eleazar Eskin,²⁷ Kevin M. Brown,¹⁸ Michael S. Marks,^{1,4,*} Stacie K. Loftus,^{20‡} William J. Pavan,^{20‡} Meredith Yeager,^{21,23‡} Stephen Chanock,^{21‡} Sarah Tishkoff^{1,2§}

Consider two geographically separated African populations; In both the skin color is reasonably uniform, but in one it is noticeably darker than the other.

If the alleles involved in the color difference between these two populations are versions of the same gene, how would you determine whether they behavior in strict dominant/recessive manner?





Where we are so far....

There are a range of interactions between alleles of the same gene: simplest co-dominance.

Blood type: A,B and O

A is dominant over O (recessive) - A antigen is present

B is dominant over O (recessive) - B antigen is present

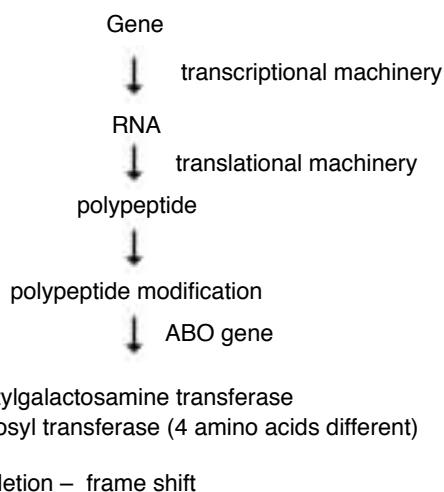
A and B are "co-dominant" - both A and B antigens are present

What does that mean.... what is the trait?

Your immune system learns to distinguish self from non-self, responds to non-self

- In the case of transfusions of blood - if you are A, you can receive A or O blood (seen as self) – B-blood leads to immune response
- if you are B, you can receive B or O (seen as self) – A-blood leads to immune response
- if you are AB, you can receive A, B, AB, or O
- if you are O, you can receive O only – A, B, or AB -blood leads to immune response

Even "simple" traits depend on many genes (genetic loci)



But, generation of A or B or AB depends upon modification of surface glycoprotein by FUT1 gene

If amorphic for fut1/fut1, there is no substrate (H-antigen) for A or B enzymes to work on, so you appear O

BUT

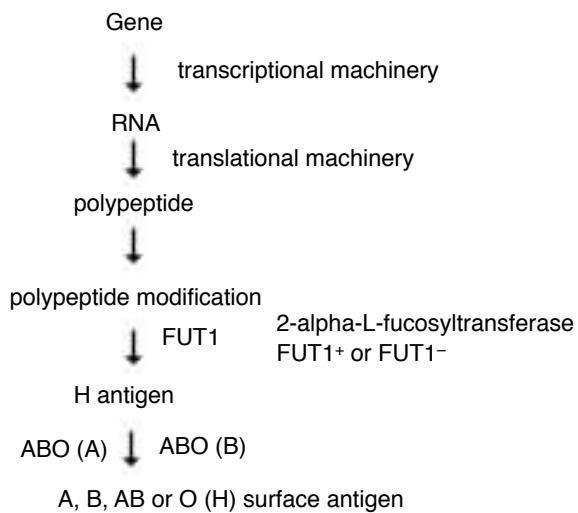
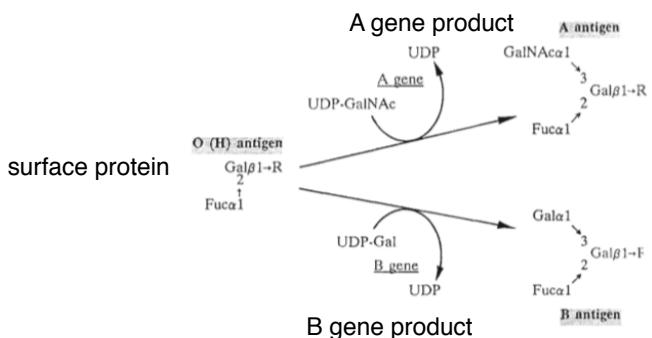
but you respond (reject) to normal O blood as foreign since normal O blood has the H-antigen (if it is FUT1+/FUT1+ it has FUT1 (H-antigen) on surface

Molecular genetic basis of the histo-blood group ABO system

NATURE · VOL 345 · 17 MAY 1990

Fumi-ichiro Yamamoto, Henrik Clausen, Thayer White, John Marken
& Sen-itiroh Hakomori

The Biomembrane Institute and University of Washington, 201 Elliott Avenue W., Seattle, Washington 98119, USA



[Genomicus of FUT1 / FUT2](#)

[EXAC FUT1/FUT2](#)