

Interactive questions

Lecture 2: Genomic variation

With the correct answers in green

Why are these blood types maintained?

- A. Reflects natural divergence
- B. Reflects ancestry
- C. Reflects climate (temperature and humidity)
- D. Reflects dietary habits
- E. Reflects disease link (infection)

Why were more SNPs reported in “Out of Africa” (Yoruban) genome?

- A. African genomes experience a greater mutation rate than e.g. Europeans ones
- B. There is more tribe isolation in Africa, hence maintenance of genetic diversity
- C. It's a consequence of human migration
- D. African genomes harbor more heterozygous sites than European or Asian ones

In the Yoruban genome, do we need to probe ...(fill in)..... SNPs than the CEU one to achieve the same threshold of ≥ 0.8 ?

- A. More
- B. Less
- C. An equal amount of
- D. I do not know

Is the average LD bin size in the Yoruban genome larger or smaller than in the European genome?

- A. Larger
- B. Smaller
- C. I honestly do not know

Using genomics to elucidate human history: *Why did Neanderthals (Ns) disappear in favor of modern humans?*

- A. They were not adapted to the changing climate
- B. Humans were more intelligent and outcompeted Ns in food collection / people care
- C. Humans are physically stronger than the “small” hobbit-like Ns and likely slaughtered most Ns
- D. Humans genetically absorbed Ns through interbreeding

Human population was larger than Ns and ultimately genetically outcompeted Ns

Some X-men mutation fun: which of the following X-men does not exist?

- A. Electric shock-proof skin
- B. Seeing 100 million color shades
(as opposed to “only” 1 million
for the average human)
- C. Immune to pain
- D. Resistance to arsenic poisoning
- E. Heat insensitive

- 1. Super Flexibility:** Those who are affected by Marfan syndrome have a mutation in the protein Fibrillin-1, which affects the body's connective tissue, giving people with this mutation the ability to bend in all different directions. Some common signs of the disorder include long arms, legs, fingers, a curved spine, flat feet, and a tall, thin body. People with the condition may also experience serious health complications affecting the heart, eyes, blood vessels, and bones.
- 2. Super Speed:** Mutations in the gene ACTN3 are associated with the ability to run fast. ACTN3 produces the protein alpha-actinin-3, which is responsible for the fast-twitch muscle fibers that allow us to run. Research has indicated that people with higher than average levels of this protein may be able to out-run the majority of us.
- 3. Resistance To Poisoning:** An entire community of people in a small town in Argentina have inherited a genetic mutation that makes them resistant to arsenic poisoning. The village of 6,000 people are known to survive after consuming more than 80 times the amount of arsenic an average person would die from. Scientists believe this phenomenon is because the village's water supply has been laced with naturally forming arsenic for thousands of years. People in this community are believed to have the gene AS3MT, which helps them flush out toxins much faster than the average person.
- 4. Resistance To Fat:** The inability to gain weight may sound like a dream come true to many people, but the reality of your body being resistant to fat is far from glamorous. MDP syndrome, which affects 8 people in the world, prevents fat from being stored under the skin. Instead, it gets deposited in the bloodstream, leading to diabetes and other health complications.
- 5. Super Vision:** A condition called tetrachromacy allows people to see almost 100 million different colors. In comparison, the average human can only differentiate between about 1 million. This is caused by a mutation in the opsin gene, which is responsible for producing visual pigments for color vision.
- 6. Shock-Proof Skin:** Slavisa Pajkic, or "The Battery Man," has a gene mutation that results in no sweat or salivary glands. Since these glands are absent, his body is able to resist electricity. According to the video, Pajkic has been known to withstand voltages as high as 20,000 volts. To put that into perspective, most people would be severely injured and burned by 50 volts.
- 7. Immunity To Pain:** Congenital insensitivity to pain (CIP) is a rare condition in which a person can't feel pain, even if they break a bone, get burned, or experience any other type of injury. CIP is caused by a mutation in the gene SCN11A. This mutation decreases the amount of sodium in the body's cells, which is key to alerting your brain of pain. CIP affects fewer than 100 people worldwide.
- 8. Super Strength:** People who have a genetic mutation in the gene MSTN will pack on muscle quickly. MSTN's job is to produce myostatin, a protein that tells the body to stop creating muscle when there's already enough. Those who have this mutation typically have at least double the amount of muscle than the average human.

Where is the "missing heritability"?

- A. Many SNPs have a very small, but relevant effect
- B. Gene X Environment But all 4 answers have some validity
- C. Structural and rare variants
- D. Genetic interactions between variants