NATIONAL CENTER FOR CASE STUDY TEACHING IN SCIENCE

Dark Skin, Blond Hair: Surprise in the Solomon Islands

by
Khadijah I. Makky and Audra A. Kramer
Department of Biomedical Sciences
Marquette University, Milwaukee, WI

Part I – Surprise

John Miller sat in his dorm room, anxious but excited to meet his roommate, Peter Koli. Peter was an exchange student from Solomon Islands who was spending the fall semester in the United States to study English.

Andrew from across the hall called out, "Yo, John, you wanna play Frisbee? We're all going."

"No thanks, I'm waiting for my roommate. He's from the Solomon Islands. I want to welcome him when he arrives."

Andrew appeared in the doorway. "Oh c'mon! It's nice out; you have all semester to welcome him. Where are these islands anyway?"

"They're part of the Melanesia Islands in the Pacific Ocean," replied John. Andrew shook his head. "OK see you later, glad to see you searched it on Google!"

When Peter arrived, he was happy to see that John was waiting for him and felt welcomed. John helped Peter get settled and they both learned a little about each other's culture.

A few days later while sitting in the park, enjoying one of the few remaining summer days, John finally asked the question that had been on his mind, "Peter, how come you have blond hair?"

"Actually, in my island it is not that uncommon to have dark skin and blond hair." Peter smiled.

"Really? That's cool! Do you know why?" John asked.

"No, I am not sure." Peter paused, then said, "I am hungry, lets go get something to eat," and he started walking towards the Student Union.

John followed. "So do your parents have the same hair color?"

"No they have darker hair, but my mom's mom has blond hair." Peter continued, "Man! I probably could have hamburgers everyday!"

"Peter, you *have* had hamburgers everyday!"

"John, I have had hamburgers much less than you have had ice cream!"

Question

1. Why do you think John was surprised to see Peter with blond hair?

Part II – Connecting Genotype to Phenotype

John was a biology student and had taken several levels of biology classes including genetics, and he started reading about skin and hair color in humans. From his reading, John learned that in humans, skin and hair color depend on the number, size and distribution of the melanin-pigment-filled melanosomes produced by dendritic cells named melanocytes. Melanin is a mixture of two pigments: eumelanin, which is black and brown, and pheomelanin, which is yellow and red. Variation in human skin and hair color depends on the variation in the ratio of these two pigments. He also learned that genetics plays a major role, since hundreds of genes are involved in the process of melanin formation. Some of these genes encode enzymes that catalyze the synthesis of the two melanin pigments, eumelanin and pheomelanin, in melanocytes. (Figure1). One of the well-studied enzymes is tyrosinase-related protein 1 (TYRP1), which catalyzes the synthesis of eumelanin. Mutations in TYRP1 have been shown to cause albinism. John also knew that all the enzymes in the pathway of melanin formation are encoded by separate genes. Alteration in the genetic code can alter the protein (enzyme) activity, leading to variations in the phenotype.

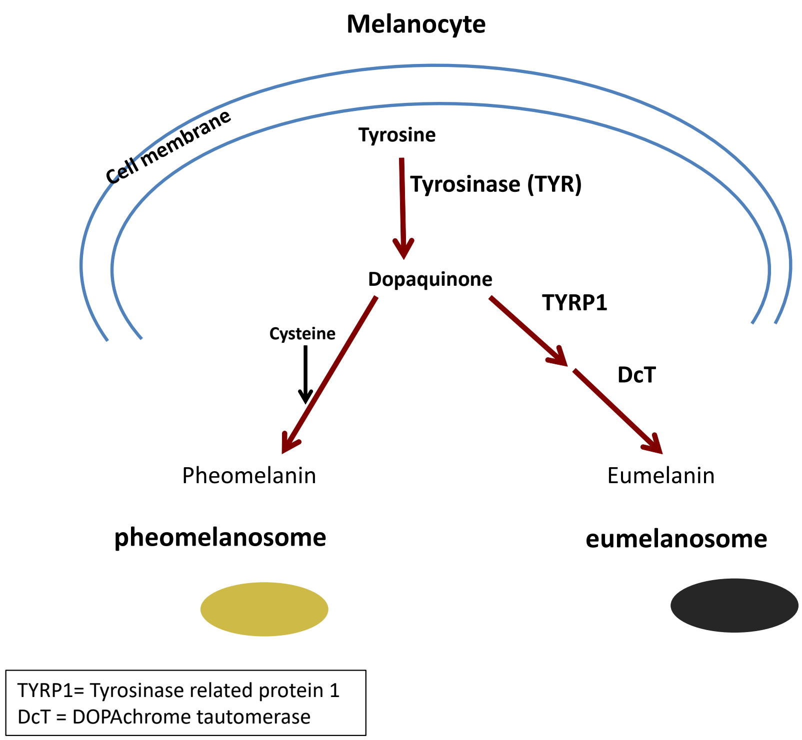

Figure 1. Melanogenesis. The schematic representation of Melanogenesis is adapted from, Hoekstra, 2006.

Question

1. What do you think would affect the production of one or both of the melanin pigments?

Part III – Genetics of Blond Hair in the Solomon Islands

A few weeks had passed and John and Peter had gotten to know each other better. "So Peter, tell me more about your islands and your people."

"What do you want to know?"

"Anything. It's an island, right? So when was it populated? Who were the first settlers? Etc."

Peter was happy to share. "The Solomon Islands are northeast of Australia in the South Pacific Ocean. They are part of a long chain of islands called Melanesia, which stretches from Papua New Guinea in the north to New Caledonia and Fiji in the south. The first human settlers came from New Guinea about 33,000 years ago. The early settlers developed land-based communities with agriculture and fishing. Europeans from Spain sporadically came in contact with the settlers on the islands starting in 1568. In the 1800s, European traders settled in the islands, and the contact with the settlers became constant. Nowadays the population of the islands is mostly made up of Melanesians; there are also some Polynesians, Micronesians, and some small pockets of Chinese and Europeans."

A few days later during his genetics class, John learned that a genetic population is a group of individuals that share a common set of genes, and that their gene pool is constituted by their shared alleles. Combinations of the alleles at different loci result in individuals with different genotypes. He also learned that populations are dynamic; the gene pool can change due to several factors such as migrations, random mating, birth and death rates. These changes can occur faster in a small population. During the lecture John started thinking about Peter and his islands.

In class John asked, "Could genetic variation due to mutations affect the gene pool of a population?" His teacher answered, "Definitely. A mutation in a locus that survives by selection generates a new allele. Propagation of the new allele will change the frequency of that allele and affect the gene pool. In fact, mutational selection just like natural selection can be a mechanism by which an allele frequency may fluctuate in a particular population."

After class John told his teacher about Peter and asked why part of the population had this characteristic and why it appeared on these islands in particular. His teacher conjectured that the population of the islands probably drifted from the larger population in New Guinea with dark skin and dark hair. The gene pool in the Solomon Islands probably used to be rich in the alleles for black hair and dark skin. A genetic variation in a hair-color-related gene occurred that generated a new blond hair allele. Because the gene pool in the islands was small, the allele frequency rapidly increased causing the appearance of this blond hair phenotype in the Solomon Islands.

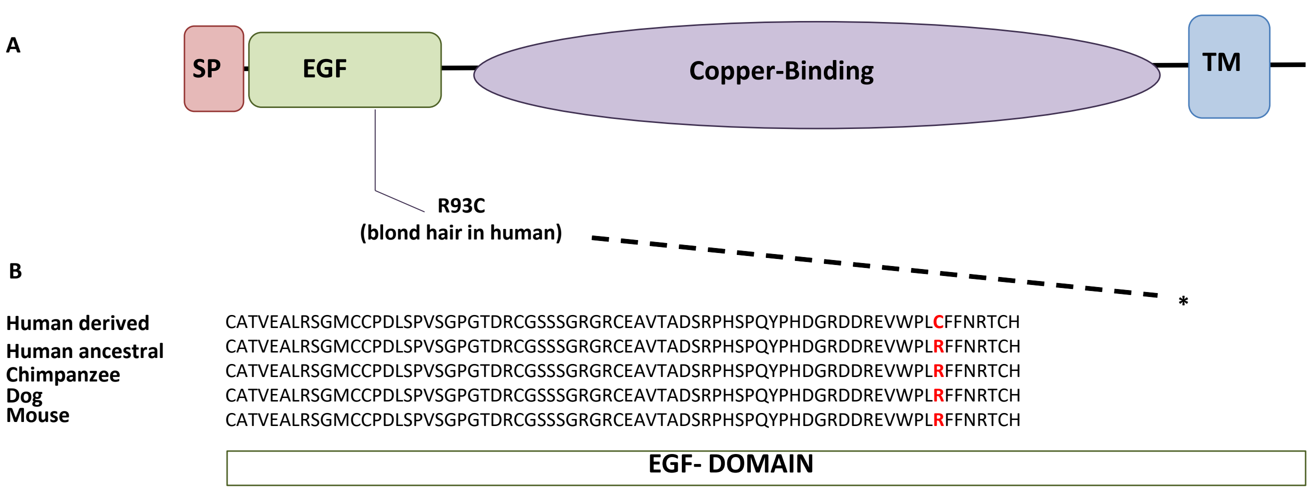

Figure 2. Schematic diagram of TYRP1 protein. A) Different functional domains of TYRP1 protein, B) Protein sequence alignment of the EGF-domain from different species and the two human variants. The figure is adapted from Kenny, 2012.

John was now even more interested. He started reading about the genomic research that was done in the Solomon Islands and came across a study that was published in 2012 (Kenny, *Science* 2012). In this study investigators showed that in the islands, roughly 5–10% of the population has dark skin and blond hair. Geneticists strongly linked a single nucleotide polymorphism (SNP) to the blond hair using genome-wide association study (GWAS) on 43 blond- and 42 dark- haired Solomon Islanders. This SNP (rs13289810) had a high frequency (0.93) in blond haired individuals. The SNP was mapped to chromosome 9 and linked to a known gene, tyrosinase-related protein 1 (TYRP1). Comparing the sequence of TYRP1 between dark- and blond- haired individuals identified a single nucleotide change C-to-T located in exon 2 of TYRP1 (Figure 2). This C-to-T transition corresponds to a change in the amino acid sequence from arginine-to-cysteine (R93C). Analysis of the protein produced from the blond hair allele revealed that this change in amino acid sequence reduces TYRP1 stability and catalytic function.

Question

1. Propose a mechanism by which the C-T nucleotide change in TYRP1 gene may have caused the blond hair trait.

Part IV – Population Genetics and Hardy-Weinberg Equilibrium

John was very excited about his findings. Later that evening he sat down for dinner and said, "Peter, I think I know the cause of the blond hair in your Islands!"

Peter smiled. "Really? So what do you think the cause is?"

"A single new mutation in the enzyme TYRP1 that reduces the dark colored pigment in the hair, so the person carrying this genetic mutation has more of the light colored pigment than the dark one—that's why the person has blond hair. This mutation occurred in the islands a long time ago. You probably got it from your grandmother who got it from her ancestors!"

Peter listened to John carefully and then said, "Nah, that is not it. My grandma always told the story of how the Spaniards and other Europeans came to the Islands and integrated into the society and mated with the natives. That led to the blond hair showing up in the islands."

John thought about Peter's story for a moment and then said, "I'm sure there's a genetic way to show which story is true."

The next day after his genetics class, John asked his teacher for help. The teacher suggested that John compare the Solomon Islands blond allele frequency among individuals in the European population to allele frequency among individuals in the Solomon Islands. John hypothesized that the islands' blond allele would have different frequencies between the two populations.

John went straight to work after class. He knew that each individual must have two alleles of TYRP1 gene. He learned that individuals with C/C and C/T alleles have dark hair and individuals with T/T alleles have blond hair. He also found that in the 2012 GWAS study, 228 individuals from Peter's islands were genotyped. The genotypes of these 228 individuals were:

# of Individuals Genotyped	Individuals with Genotype C/C	Individuals with Genotype C/T	Individuals with Genotype T/T
228	126	80	22

The researchers in the 2012 GWAS also looked at the distribution of the Solomon Islands' blond allele around the world. They genotyped 944 individuals from 52 populations (different countries) with the following results:

# of Individuals Genotyped	Individuals with Genotype C/C	Individuals with Genotype C/T	Individuals with Genotype T/T
944	941	0	3

John hurried back to the dorm, confident that he now had the information that would convince Peter about the origins of his blond hair.

Questions

1. Using the Hardy-Weinberg equilibrium:
   a. What are the observed genotype frequencies in the Solomon Islands?
   b. Calculate the blond hair allele frequency and the dark hair frequency in the Solomon Islands and worldwide.
   c. If we assume that the population follows Hardy-Weinberg assumptions, what is the expected frequency of heterozygous carriers in both populations?
2. How do you think the gene for blond hair was introduced to the islands?

References

Kenny, E.E., N.J. Timpson, M. Sikora, M.C. Yee, A. Moreno-Estrada, C. Eng, S. Huntsman, E.G. Burchard, M. Stoneking, C.D. Bustamante, and S. Myles. 2012. Melanesian blond hair is caused by an amino acid change in TYRP1. *Science* 336(6081): 554–561.

Hoekstra, H.E. 2006. Genetics, development and evolution of adaptive pigmentation in vertebrates. *Heredity* 97: 222–234.

Loury, E. 2012. Blond afro gene study suggests hair color trait evolved at least twice. *The Huffington Post* 05/06/2012. <http://www.huffingtonpost.com/2012/05/04/blond-afros-melanesia-genetic-inheritance-independent_n_1479160.html>.

McCarthy, M.I., G.R. Abecasis, L.R. Cardon, D.B. Goldstein, J. Little, J.P.A. Ioannidis, and J.N. Hirschhorn. 2008. Genome-wide association studies for complex traits: consensus, uncertainty and challenges. *Nature Reviews Genetics* 9: 356–369.

World Culture Encyclopedia: Countries and their Cultures [Website]. *n.d.* Solomon Islands. <http://www.everyculture.com/Sa-Th/Solomon-Islands.html#b>.