

Albinism Advanced: Across the Species

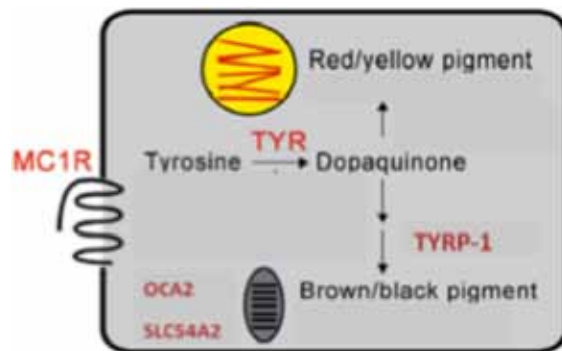
Oculocutaneous albinism is a group of conditions that affect coloring (pigmentation) of the skin, hair, and eyes. Approximately 1 in 20,000 exhibits this disorder and affected individuals typically have very fair skin and white or light-colored hair. Melanin is an extremely important molecule in humans for many reasons, one being its ability to protect the DNA in the cell's nucleus from damage by UV light from the sun. This is one reason the skin darkens after being exposed to sunlight and people living closer to the equator have darker skin for this extended protection.

Albinism is actually found across all major animal groups since nearly all produce melanin. It has been observed in mammals, birds, reptiles, amphibians, fish and even invertebrates. This is not surprising since the genes for melanin production, the proteins responsible for producing pigment in skin, hair and eyes, are similar among these species. All it takes is one small change to result in a change in a protein's shape to make it non-functional. (NCBI; www.ncbi.nlm.nih.gov)

What genes contribute to different hair/fur colors in mammals?

The genetics of coat color in animals includes many genes in a complex biosynthetic pathway. The amino acid tyrosine is first converted to dopaquinone by the enzyme TYR, for tyrosinase, and will be designated as the "C" gene (for "color"). If tyrosinase is not present, the animal will be albino.

Dopaquinone can be converted to both phaeomelanins which are yellow/red pigments, or eumelanins, which are brown/black pigments. In the eumelanin pathway, dopaquinone first is converted to a brown pigment. The enzyme TYRP-1 converts brown pigment into black, and will be designated as the "B" gene. Mice that don't have a functional TYRP-1 gene will be brown and have the genotype "bb". The MC1R receptor is the receptor for the hormone MSH (melanocyte-stimulating hormone), and will be designated as the "R" gene (for "receptor"). When MSH locks onto the MC1R receptor, this signals the inside of the cell to convert dopaquinone to eumelanin at the expense of phaeomelanin. If the gene for the receptor is mutated, i.e., "rr", dopaquinone can still be converted to phaeomelanins, leading to yellow (blond) or red hair/fur.



This activity will focus on only a few of the genes necessary for pigment production. However, you should be aware that there are many more genes that contribute to hair/fur and skin color; those mentioned above and others that have not been mentioned.

Part One: Black, brown and white mice (dihybrid cross with a “twist”)

In this part of the activity, we will focus on just 2 genes: tyrosinase or “C”, and TYRP-1, or “B”.

1.) A laboratory researcher mated a black mouse to a brown mouse. The F1 mice were all black. What is the genotype of the F1 mice? (Hint: remember what the C gene codes for-what would happen if the mice were “cc”?)

2.) The researcher mated two of the black F1 mice. Do a dihybrid Punnett Square to determine the phenotypic ratio (fur color) of the F2 mice.

3.) A dog breeder wanted to start breeding dogs to sell yellow labs. He had a yellow male and a chocolate female. What are the possible genotypes of a yellow lab for the R gene and the B gene? What will the F1 puppies look like? When the F1’s were born, he was very surprised! Why?



4.) The breeder took two of the F1 puppies and mated them. Do a dihybrid Punnett Square to determine the phenotypic ratio (fur color) of the F2 puppies.

- 5.) Zebrafish are named because of the dark stripes running along their sides. The dark stripes are due to the pigment melanin, which is the same pigment responsible for hair, eye, and fur color in mammals. A few zebrafish, however, have been found that have light stripes and are called “golden” fish. Scientists know that this trait is genetic because golden fish have golden offspring. The gene responsible for this change is SLC24A5. Replacing the fish SLC24A5 gene with the human version restores the black stripes in the zebrafish. How is this possible?



a zebrafish with the
“golden” mutation



a zebrafish as it is
found in the wild

- 6.) Researchers have determined that SLC24A5 mutations may only account for 25-30% of differences in skin color in humans. Why?

- 7.) A male albino recessive for tyrosinase and a female albino recessive for SLC24A5 marry and have a baby. To their surprise, the baby is not an albino. Why?
