

Blue-eyed individuals

Maxime Coles MD

I recently was examining patients in an orthopedic clinic in Naples, FL when one patient attracted my attention. I was stunned at the sight of one patient, a blue-eyed gentleman from our country of Haiti. For someone who travelled so extensively, in all the corners of our island, during my medical missions, it felt in me an urge to investigate the part of the island he was coming from. I became curious to ask for his place of birth. A nice and healthy 35-year-old gentleman, manual worker, from Bainet, Haiti. I have to say that I met so many of patients from Fonds des Blancs or Casale or Fort-Liberty Haiti with such characteristic eye colors, but this one made the most effect of surprise on me, especially perhaps because, I was in the middle of nowhere in Naples FL I was not expecting such surprise and I gave him all my attention.

“Bainet” has a multiracial population of African and European descent in our country of Haiti which remind a little, other cities like “Casale” and “Fonds des Blancs” which are home to an exceptional and typical ethnic group that sometimes because of their beauty, they over the years, called “Marabouts”. The inhabitants of Bainet, more than anywhere else in the country. describe the aspect that I have tried to described earlier in the patient described above. Often, they may have European features like curly or wavy hairs, or even straight hair, on a dark skin. The eyes are beautiful varying from a pale to a dark green, or to a sky-bleu or a hazel color with represent a mixture of brown, or gold added to the sky-bleu. Etymologically, these population have existed in the country, from the time of our war of independence when many soldiers from the Napoleon Army, joined the Haitian revolution to fight the same French army who brought them on Santo Domingo soil to re-establish slavery. The population of those exotic town of Haiti have given a mixture of French, Middle East, African descents, especially Polish, of an extreme beauty which over the years have taken different names, notably in Bainet, the term “marabout” has been used to describe such elegance and beauty.

He was working at a packing facility, for a factory, enjoying his daily shifts of activities. He did not wear any glasses although he was suffering from a mildly restricted vision and was diagnosed with early stages of cataract. He was told that at maturity, he will need an operation to implant lens and restore his vision. The pupils appear irregular with an advanced cataract noted with the opacified lens. The Iris has a display of color where a sky-blue color was dominant tinted with brown and gold color, in a rare display of colors. where the clear sky-blue was predominant, tinted with patches of brown as well. He was not wearing any glasses but was told that in a near future, he may require a surgical intervention. He allowed me to photograph his eyes and display the photographs. but the lighting did not permit me to render the beautiful shade of color Nature doted him with. He admitted being the only member of the family with the characteristics of blue-eyed but recalled his father with the same curious eyes color when he was alive.

Eye color (colour) is generally modeled as a Mendelian trait but recent researches of inheritance have also demonstrated that eye color does not follow the classical path. At least 16 different genes have been proven to be responsible for eye color, but two proteins (HRC2) and another one responsible for oculocutaneous albinism (OCA2) have been found associated to the chromosome 15. An “intron” in HRC2 promote the OCA2 expression. It looks like one single nucleotide in any of these two genes seems to play a more predominant role in the eye colour. Some individual may express different phenotypes in each eye with perhaps, sometimes, a complete absence of pigment in one eye.

This is a little why, “Eye-color” is often used to teach Genetics to explain the basics of the Mendelian genes as a recessive or dominant or X-lined disorders. Beyond Genetics, color perceptions touch on other concepts involving optics, physics, psychology and physiology, has captured the imagination of the population. Eye-color is determined by the variations in a person’s genes. Most of the genes associated with eye-color are involved in the production, transport or storage of a special pigment called “Melanin”. The allele genes come in the form of brown, blue or green where the brown is the most dominant.



Hazel - Sky blue eyes

Bainet is very multiracial, but the majority of Bainet's population are of European and African descent. Bainet was found to be the home to an exotic Haitian ethnic group called “marabouts”. Marabout are people with dark skin with beautiful blue, green or hazel eyes or of dark skin with European features, straight, curly or wavy hairs. They are a mix of Native, French European, African, and Arabic descents. The name of “Marabout” was given to this original and ethnic group endemic to Bainet, Haiti.

Commune in the arrondissement of Bainet, in the Sud-Est department of Haiti, the town of Bainet is multiracial, but the majority of the population is of European and African descents with either dark skin with gorgeous blue, green or hazel eyes or fair skin with European features like straight, wavy or curly hair. Less than 150,000 inhabitants live in this corner of the island. It is the place where apparently, you can find the friendliest persons in the island.

Scientists know now, how to use a complex genetic trait to determine the specific roles of different genes. We have learned that these genes control the amount of melanin available in the specialized cells, allowing the determination of the eye-color. Science has reached the conclusions that the pigment “melanin” is a yellow-brown pigment in the body, notoriously able to determine the skin color and the

color of the iris. It is a family of molecules that act like a pigment in many organisms, including humans. It is the most common light-absorbing in living organisms and is generally brown or black in color.

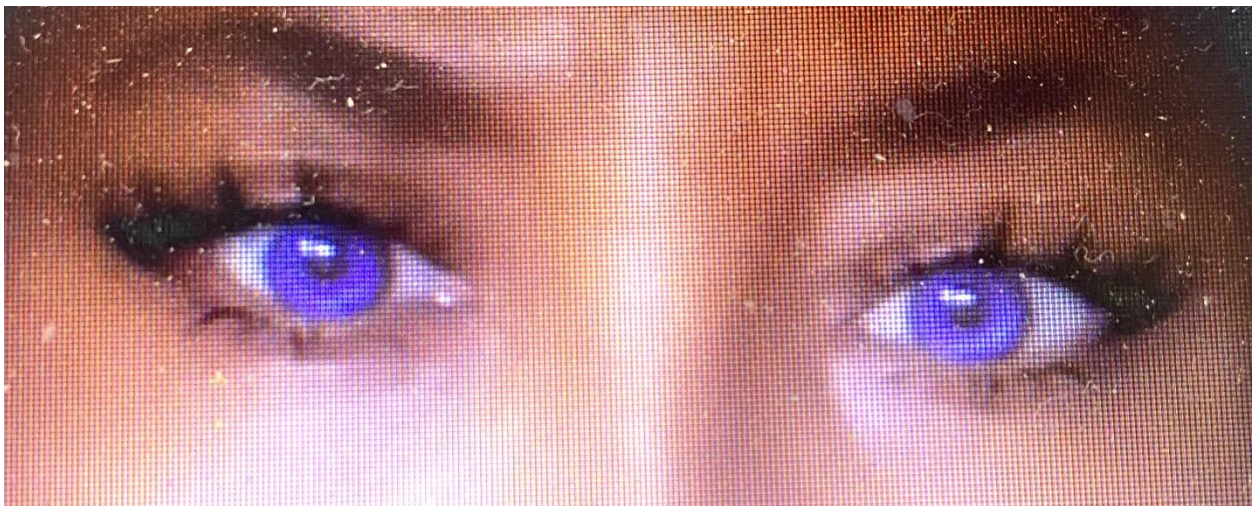
Melanin is a black pigment synthesized from Dopamine. L-Dopa and L-tyroxine. Melanin containing cells including catecholaminergic cells in the brain and melanocytes in the hairs, and skin, pigments cells of the inner ear, iris, and choroid of the eye. The function of melanin is quite different in the neural versus peripheral cells. Melanin pigment is the basal layer of the epidermis and can absorb most of radiation from the sun to provide a protective effect.

Three types of Melanin were determined:

1-Eumelanin, a black-brown responsible for the intensity of the dark eye, composed of indole derivatives produced by auto-oxidation of Dopamine. It is the most abundant melanin in the body and the predominant melanin pigment in hair and the skin.

2-Pheomelanin, a red-orange pigment found in the eye of people with green and hazel eyes contains benzetimide molecules from incorporated cysteine or GSH with dopamine-quinone derived by dopamine by auto-oxidation. Pheomelanin (Phaeomelanin) is more prominent in redhaired or blonde-haired individuals. The Melanosomes carry a mixture of the two pigments.

3- Melanin can be produced by a variety of organisms including fungi and have been used in the development of novel therapies.





Beautiful Sky blue eyes in three young Haitian girls

Researchers at the University of Copenhagen (2008) have demonstrated that people with the characteristic “blue-eyes” have a single and common ancestor. A

gene mutation has been traced around 10,000 years ago and it is believed to be the cause of the all blue-eye color among humans presently alive on the planet. One region of the chromosome 15 contains two genes (OCA2 and HERC2) and both play a role in the color of the eyes.

Professor Hans Eiberg at the Department of Cellular and Molecular Medicine stipulated that all individuals originally had brown-eye but is believed that a gene mutation is responsible for the change of color. The OCA2 gene' on our chromosome 15, appears to be responsible for this change of color from the brown to the blue. This gene produces a "P-Protein" which help the melanosomes mature and store the melatonin in the iris. The OCA2 gene controls almost three-fourths (3/4) which is responsible of the production of the melanin pigment, responsible for the color to our eyes (blue-brown spectrum), hair and skin. The gene adjacent to the OCA2 (HEC2) limits its action in reducing the production of the melanin in the iris, and diluting the brown eye color into blue, compared to the possibility that the protein OCA2 being completely destroyed or turned off. This will result in an absence of melanin in the iris, eyes and skin like we see in peoples with albinism. If both parents have already blue-eye, there will be almost 99% of chances that the child will present with blue-eyed iris and 0% chances to have a kid with brown eye. It seems that some studies may have shown that almost 16 different genes can be responsible of the eye color in humans.



The rarest eye color is green in 9% of American and only 2% in the world population. Some may say that the grey eye color is the rarest nowadays. The other rarest is the hazel/amber color.

The amount of melanin explains the variation of color in the eye (iris) from brown to blue but the blue eye individuals have only minute amount of melanin in their eyes. This is the way; the scientists have concluded that any blue-eyed person is linked to the same old ancestor as reported by Professor Elberg because they have inherited the same switch on their DNA. There have also limited genetic variations in the color of the eyes from brown to green which can all be explained by the amount of melanin in the iris. They have all inherited the same switch at exactly the same spot in their DNA but they have more variation in the area of DNA controlling the production of melanin. Even the mitochondrial DNA was studied for the blue-eye color individuals in countries like Jordan, Denmark, Turkey in 1996 by Professor Eiberg who also explained that mother Nature is constantly changing the human genome and mixing chromosomes. The allele for the brown eye is the most dominant allele by dominant over the other two alleles, while the allele for green-eyed is always dominant over the allele for blue-eyed which is also recessive.



It may be possible for two parents with brown-eyed color with each passing a blue-eyed allele to an offspring for the child to heritage a blue-eyes at birth or if the parents transfer a green-eyed allele to the offspring, the child may have grey-eyed but if each parent passes a brown-eyed allele to the offspring, the child will have brown eyes. The allele brown being a dominant allele. The more melanin you have, the darker will be your eye. Black is not an eye color. The rarest eye color used to be green (2%) worldwide but it seems that nowadays, the gray color has become the rarest color. He hazel-amber color

is the next rarest color of the eyes. Everyone with blue-eye may descend from a single human ancestor.

There is also a group of genetic conditions causing hearing loss and changes in pigmentation of the skin, eyes and hair classified under the name of “Waardenburg syndrome” in which there is a progressive loss of hearing to both ears as well as an association with diseases of the hands and malformations or association with intestinal disorders like Hirschsprung syndrome with different degrees of intestinal obstruction, and constipation etc. I will not extend much deeply on this pathology, but people suffering from that disease, have very pale blue eyes or even eyes with different colors such as one is blue and the other is brown. There are fewer than 50,000 people in the United States with this disease and the symptoms generally appears at birth or I then infancy. Waardenburg syndrome (disease) is caused by a change in the DNA and is seen among Kenyan Africans and around 5% of these children will develop a congenital deafness. We would like to note that such disease was described as well in the population of Northern Europe. It becomes interesting that I chose to treat this subject for the AMHE Newsletter because we are about to receive a delegation of Kenyan soldiers to pacify our country.

Finally, for the purpose of discussion and to enlighten our medical students and the physicians in training, I wanted to open a parenthesis to describe also a viral disease manifested by “blue eye”, which is caused by a porcine virus (La Piedad Michoacan Mexico virus) “Porcine orthorubula virus” commonly identified and causing encephalitis, pneumonia and respiratory disease in little pigs (piglets). It attacks also the reproductive system of the adult pigs, rendering them infertile. The presence of a corneal opacity and scarring is described with neurological symptoms like weakness, ataxia, muscle tremors and abnormal posture with rigidity of the legs and respiratory problems. The virus is transmitted directly or indirectly by fomites and birds, it is only at autopsy that a diagnosis can be made on the dead animal. It was identified in the 80’s, in Mexico.

The virus responsible of this disease is an RNA virus similar to Measles, Mumps, and Rubella or para-influenza or respiratory syncytial virus seen, in humans. It affects rarely humans in close contact with the infected animals or at proximity.

Maxime Coles MD
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