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# Phenotypic Variation in Pigmentation of Persons with Albinism in Rejang Lebong, Bengkulu

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**ABSTRACT**

Albinism is a congenital disorder, a group of genetic disorders in which there is a partial or total lack of the pigment melanin in the eyes, skin, and hair. It is caused by one or more enzymes committed to the biosynthesis of melanin, and one of them are a deficiency of the tyrosinase. It is a genetic disorder transmitted by an autosomal recessive gene. The research was conducted from August-November 2019. It was aimed to determine the variety of pigmentation on the skin, hair, and eyes of the human population with albinism disorder in Rejang Lebong, Bengkulu Province, by using the descriptive research method based on qualitative data. Data were collected by interviews with the family of persons with albinism disorder. The results showed that the color of skin, hair, and eyes are varied between individuals, although they are siblings. The skin color is varied from white to reddish or pale brown. Hair color ranges from fawn, brown, reddish-yellow to reddish-brown, while the color of the eyes is more varied, from yellowish-brown, dark-brown, light-grey, greenish-blue, bluish-black. About 57% of the participants had strabismus problems, and 43% had iris transillumination,1 00% had nystagmus. They all come from the family that had phenotypically normal parents. More than 50% of the family members did not have the albinism phenotype until the previous 2-3 generations.

***Keywords**: albinism, Bengkulu, pigmentation*

## 1. INTRODUCTION

The pigment is a substance that gives color to the human body, animals and plants. Due to the presence of pigments, human skin color varies, from the darkest brown to the lightest. The color of skin, hair and eyes is determined by the presence of the pigment melanin. Melanin is a pigment that is responsible for the appearance of color in the hair, skin and eye [1]. Melanin has an important role in preventing DNA damage caused by sun exposure. In general, there are two kinds of melanin, namely eumelanin and pheomelanin. Eumelanin is responsible for dark pigments (dark brown to black) and pheomelanin is responsible for light colors (yellow, red, and blonde) [2]. Melanin is produced in melanocyte cells. Melanocyte cells are derived from melanoblast. Failure to form melanin is caused by damage to the enzyme Tyrosinase. Tyrosinase enzyme is encoded by the tyr gene. It is involved in the process of melanin formation [3].

Failure to synthesize melanin can lead to albinism characterized by the absence of pigment or less melanin pigment in hair, eyes and skin [4]. Based on the morphology, albinism is grouped into two major groups, namely Oculocutaneous Albinism (OCA) and Ocular Albinism (OA). OCA has symptoms of lack or absence of pigment in the skin, hair, and eyes. Meanwhile, OA has symptoms of the absence of pigment only in the eyeball. According to Newton *et al*. [5], Oculocutaneous Albinism (OCA) is divided into several types, namely OCAIA, OCAIB, OCA2, OCA3, and OCA4. Meanwhile, Ocular Albinism (OA) can be divided into 3 types, namely OA1, OA2, and OA3. Research on pigmentation variations of albinism has been carried out in Kedurang, Bengkulu Selatan District. It was concluded that the albinism pigmentation in Kedurang had different degrees of

phenotype variation, resulting in skin and hair pigmentation changed. The melanin production increase during childhood to teenager. The types of albinism in Kedurang District are OCA1, OCA2, and OCA3. The results are obtained from morphological data or phenotypic observations [2]. There are some people with albinism disorder in the Rejang Lebong, Bengkulu. As their pigmentation variation in skin, hair, and eyes have not been investigated, we carried this research to collect the data based on phenotypic variations in pigmentation hair color, eyebrows, eyelashes, iris, strabismus, nystagmus, and iris transillumination.

## 2. MATERIALS AND METHODS

The research was conducted to the family with albinism disorder in Rejang Lebong Bengkulu Province. It was aimed to determine the variety of pigmentation on the skin, hair, and eyes of persons, by using the descriptive research method based on qualitative data. The data were collected by interviews with the family of persons with albinism disorder. Color determination refers to the Munsell Color Index, by Android-based mobile application and The Munsell Color Tree [6]. The steps to determine the color using the Munsell Color Index are:

- The surface of the skin/hair/part of the eye be under sufficient light
- Munsell Color Index is brought closer to the surface of the skin/hair/eyes
- The observed color is adjusted to the closest Munsell Color

Observation of the determination of pigmentation variations in persons with albinism in Rejang Lebong Regency uses the Munsell color tree [6]. Data was collected from persons with albinism disorder who had agreed and signed a statement of willingness to become a volunteer.

## 3. RESULT AND DISCUSSION

The determination of pigmentation variations of persons with albinism was carried out based on the phenotype of hair color, eye brows, eyelashes, exposed and unexposed area of skin, iris, as the following table. All the participants had pigmentation variations in hair, skin, and eyes. Variations were also seen in participants with albinism who were closely related (siblings). This variation seems not only caused by genetics but also caused by the duration spent outdoor and exposure to sunlight every day. Participants who have activities outdoor and are exposed to more sunlight have reddish skin tones, while those who are more indoor, protected by sunlight, tend to have white skin tones without redness. Based on King [7], the reddish pigment is caused by sunburn so that the color of the blood pigment rises up but the melanin pigment remains undeveloped. A fairly high pigmentation variation also occurs in a family with albinism disorder in Kedurang Bengkulu [8]. Three people with albinism who are sibling are classified as type OCA2 (the elder child), type OCA3 (the middle child), and type OCA1b (the youngest).

**Table 1**. Observation of the phenotypic color of hair, eyebrows, eyelashes, skin and iris of persons with albinism in Rejang Lebong Regency Bengkulu

| Age group | Color | | | | | | |
|---|---|---|---|---|---|---|---|
| | **Hair** | **Eyebrow** | **Eyelash** | **Sunlight-unexposed skin** | **Sunlight-exposed skin** | **Outer iris** | **Inner iris** |
| Underage (9-16 years old) | fawn, brown | white, pale-yellow, reddish-yellow | white, pale yellow, reddish-yellow | white, reddish-white, pink | reddish-white, pink | greenish-blue, yellowish-brown | bluish-black, dark-brown |
| Youth (20-47 years old) | reddish-yellow, brownish-yellow | pale yellow, reddish-yellow | pale-yellow, reddish-yellow | reddish-white, pink | reddish-yellow, pale brown, pink | greenish-blue, yellowish-brown, light-gray | bluish-black, dark-brown |

Age group based on WHO classification, 2020

Although it still needs to be confirmed, it is suggested that people with albinism in Rejang Lebong do not change in pigmentation as happened in the albinism family in Kedurang, South Bengkulu. In the family, there are three people with albinism disorder. Their pigmentation increased in the skin, hair, and eyes during their childhood to teenage. There is an improvement in their eyes. As they got older, they have no nystagmus and strabismus. The increasing of pigmentation affects the resistance of each individual to sun exposure [8], [9]. According to King et al. [10], a significant increase in pigmentation could occur because there is still 1-10% tyrosinase activity. Besides, outdoor activity can also be the trigger to the development of pigmentation in the body.

All of the participants with albinism disorder from Rejang Lebong had eye problems. This problem happens both in the underage group and the youth. As many as 57% of the participants had strabismus problems, and 43% had iris transillumination. This condition causes people with albinism to get amblyopia and photophobia [11], [12].

Based on the study, hair color in people with albinism are varied, ranging from fawn, brown, brownish-yellow, reddish-yellow. While normal people in this family have the darkest hair colors, ranging from dark brown to black. Based on the phenotypic characteristics of hair, it was estimated that people with albinism in Rejang Lebong Bengkulu tended to be OCA2 and OCA3. According to Manga [13], albinism type OCA2 has hair colors ranging from brown, ginger/ginger to reddish blonde. This is due to mutations in the P gene that codes for the P protein. It is caused the develop of pheomelanin, resulting yellow or red pigments. Albinism type OCA3 [7] has hair colors ranging from red to red-brown. However, molecular methods are needed to determine the types of this albinism.

The eyebrows and eyelashes have similar color variations. The colors range from white, pale yellow, reddish-yellow. While the eyebrows and eyelashes of other normal siblings have colors ranging from dark brown to black. According to King [7], persons with albinism have pale eyebrows and eyelashes that indicate a null mutation. There is no melanin pigment. In the meanwhile, dark color indicates the presence of a leaky mutation type, especially OCA1b type albinism, so that some albinos still produce pigments. In previous studies [14], people with albinism in Kaur Regency Bengkulu have different pigmentation of eyebrows with a spectrum range from yellow, pale yellow, brown. dark, and very dark grayish brown. Meanwhile, normal people have black eyebrows. The color of the eyebrows and eyelashes of people with albinism range from yellow to red indicates the formation of a small amount of pheomelanin. Meanwhile, the color of the eyebrows and eyelashes of normal people who have a brown to black color indicates the presence of eumelanin [4].

Based on tracing family tree through interviews, all participants come from families where both parents have normal phenotypes, there are no pigmentation problems. More than 50% of the family members did not have the albinism phenotype until the previous 2-3 generations. The phenotype of albinism does not only appear in families that have a marriage line from related individuals based on the pedigree diagram, but also appear in families who are not closely related (come from different tribes).

Variations of skin color covered by clothes are paler than exposed skin, varies from white or pink to reddish or and pale brown. If the skin color remains pale even when exposed to sunlight, this means that this type is categorized as albinism null mutation. When it is exposed to sunlight, the albinism null mutation type will show reddish pigmentation. Null mutations in tyrosinase occur due to changes in DNA bases contained in codon number 81 so that tyrosinase activity fails [2].

Outer iris pigmentation of people with albinism is not common among the ethnic groups in Bengkulu. The color of the outer iris varies from greenish-blue, yellowish-brown, light gray, while the inner iris is bluish-black or dark brown. The common color in other family members with normal phenotypes is dark brown or blackish-brown. This color is commonly found in the tribes in Bengkulu. The color of the outer iris of the eye is lighter than the inner iris, both in the persons with albinism and normal family members. Other research carried to group albinism in Kaur Bengkulu [14], found that the outer iris color of persons with albinism are light gray, brown, while normal people have a black or dark brown. There is greenish-blue color in the outer iris. According to [7], [13], OCA1 type albinism has a reddish-brown iris. Albinism type OCA2 has a blue-green iris. Meanwhile, OCA3 type albinism has a blue or a brown iris. The greenish-blue iris indicates that the people with albinism in Rejang Lebong, Bengkulu are group of albinism type OCA2 or OCA 3.

## 4. CONCLUSION

In conclusion, based on the phenotypic variation of pigmentation of hair, eyebrows, eyelashes, skin, iris, persons with albinism in Rejang Lebong Bengkulu are suggested a group of albinisms OCA2 and OCA3. About 57% of the participants had strabismus problems, and 43% had iris transillumination, 100% had nystagmus. All participants were born to families with phenotypically normal fathers and mothers. More than 50% of the family members did not have the albinism phenotype until the previous 2-3 generations. The phenotype of albinism does not only appear in families that have a marriage line from people that are closely related but also in families who are not closely related (come from different tribes).

## REFERENCES

[1] C. Muslim, B. Karyadi, B. Suryobroto, Penentuan tipe albinisme di Kecamatan Kedurang Bengkulu Selatan dengan sidik

RFLP-PCR, FMIPA Universitas Bengkulu, 2007 [In Bahasa Indonesia]

[2] C. Muslim, Genetika warna. UNIB Press, Bengkulu, 2008 [In Bahasa Indonesia]

[3] C. Dessinioti, A.J. Stratigos, D. Rigopoulos, A.D. Katsambas, Sebuah ulasan tentang kelainan genetik hipopigmentasi: pelajaran dari biologi melanosit, Dermatologi Eksperimental. Vol. 18(9) (2009) 741-749 [In Bahasa Indonesia]

[4] D.P. Snustad, M.J. Simmons, J.B. Jenkins, Principles of genetics, Wiley, New York USA, 1997

[5] J.M. Newton, O. Cohen-Barak, N. Hagiwara, J.M. Gardner, M.T. Davisson, R.A. King, Mutation in human orthologue of the mouse underwhite gene (uw) underlie a new form of oculocutaneous albinism OCA4, Am. J. Hum. Gen, 69 (2011) 981-988

[6] A. Munsell, The Munsell Color Tree. https://munsell.com/color-blog/color-tree/, (accessed January 20th 2020), 2020

[7] R.A. King, C.G. Summers J.W. Haefemeyer, B. LeRoy, Facts about albinism international albinism Center at The University of Minnesota. USA, 2000

[8] E. Puspita, Variasi pigmentasi albinisme di Kecamatan Kedurang Bengkulu Selatan, FKIP Universitas Bengkulu, 2003 [In Bahasa Indonesia]

[9] Sipriyadi, Perkembangan fenotip dan penentuan genotip RFLP-PCR (Restriction Fragment Length Polymorphism-Polymerase Chain Reaction) pada satu keluarga albinisme di Kecamatan Kedurang Bengkulu Selatan, Konservasi Hayati Vol. 05(01) (2009) 55-67 [In Bahasa Indonesia]

[10] R.A. King, C.G. Summers, J.W. Haefemers, B. Leroy, Fact about albinism. International Albinism Center at the University of Minnisota, USA, 2001

[11] A.M. Ansons, H. Davis, Diagnosis and Management of Ocular Motility Disorders. (4th Ed), Wiley-Blackwell, 2014

[12] C.G. Summers, 2009. Albinism: Classification, Clinical Characteristics, and Recent Findings. Optometry and Vision Science, 86(6) (2009) 659–662

[13] P. Manga, J. Kromberg, A. Turner, T. Jenkins, M. Ramsay, in southern Africa, brown oculocutaneous albinism (BOCA) maps to the OCA2 locus on chromosome 15q: P-gene mutations identified, Am J Hum Genet 68(3) (2001) 782-787

[14] D.P. Lestari, Sidik RFLP-PCR dan SSCP pada ekson 3, 4 dam 5 gen tyrosinase terhadap keluarga albinisme di Kabupaten Kaur dan Kota Bengkulu. FMIPA Universitas Bengkulu, 2008 [In Bahasa Indonesia]