A Cross Sectional Survey of the Knowledge and Attitude Towards Albinism Diseases and Prevalence of Albino in District Khairpur

Majeeda Ruk
Department of Zoology, Shah Abdul Latif University Khairpur, Sindh Pakistan

Javed Ahmed Ujan
Department of Zoology, Shah Abdul Latif University Khairpur, Sindh Pakistan
Postdoc Fellow at the Department of Animal Sciences, (IFAS), University of Florida, USA, javed.ujan@fulbrightmail.org

Khalida Unar
Institute of Microbiology, Shah Abdul Latif University, Khairpur

Shazia Parveeen
Department of Biochemistry, Shah Abdul Latif University, Khaipur

Aiman Amur
Department of Zoology, University of Sindh Jamshoro

Asif Ali Soomro
College of Pharmacy, Liaquat University of Medical and Health Sciences Jamshoro

Ayaz Ali Unar
Institute of Pharmacy, Shaheed Mohatarma Benazir Bhutto Medical University, Larkana

Shah Noor Suhriani
Government Islamia Arts & Commerce College, Sukkur

Shafi M. Wassan
Department of Community Medicine, GMMMC, Sukkur

Sham Lal
Institute of Microbiology, Shah Abdul Latif University, Khairpur

Abstract

When a person's skin, hair, or eyes are abnormally light in color, we use the clinical word "albinism" to refer to their condition. Mutations in genes involved in melanin production, melanocyte differentiation/migration, or membrane trafficking cause this condition. Albinism is one of those disorders

that tends to be very uncommon overall, but the founder effect has caused it to become more common in certain areas than others. Albinism (sometimes known as "albino") is a condition passed down from parents to kids due to a lack of melanin. Those 5-year-old are most at risk. Typically, these illnesses invade our body (brown hair, white boy blue or green eyes). Ocular albinism type 1 (OCA1) is characterized by abnormally light retinal and iris pigmentation. Skin and hair color are unaffected by this disorder. Complete lack of melanin in the hair and skin is characteristic of oculocutaneous albinism type 2. The study's primary goal was to gather secondary data from several locations in District Khairpur in order to compare albino patients across demographic categories such as gender, age, and tehsil. Between July 2022 and November 2022, 20 people were reported to have albino illnesses. The patient's knowledge and outlook were analyzed using secondary data, with frequency and percentage counts taken and the results described using bar charts and pie charts, respectively. Generally, the ages ranged from ten to thirty. Among the total of 20 recorded cases, 4 were male (20%) and 3 were female (15%). In the district of KhairPur, 13 of the instances involved children. Albinos, their families, the general public, educators, medical professionals, and governmental agencies may all benefit from learning more about albinism via health education campaigns.

Keywords: *Albinism, Khairpur, OCA, Attitude, Knowledge.*

Introduction

The word "albinism" refers to a broad category of clinical diseases marked by the hypopigmentation of a person's skin, hair, and eyes, or only their eyes. It is brought on by pathologic variations of genes involved in membrane trafficking, melanocyte differentiation/migration, or melanin production (Bastonini et al., 2019). There are two varieties of albinism: syndromic albinism, which manifests a variety of non-pigmentary symptoms such bleeding diathesis, lung fibrosis, and immunodeficiency, and non-syndromic albinism, which only manifests symptoms of defective melanin manufacture. We can now identify more kinds of albinism and the genes that cause these conditions thanks to recent developments in genetic research. Here, we update and examine the pathogenesis of albinism as well as its clinical characteristics.

Albinism is an autosomal recessive condition brought on by a reduction in melanin formation in melanocytes or its entire absence (Marçon & Maia, 2019). Albinos are more vulnerable to the damaging effects of UV light and are more likely to develop actinic damage and skin cancer because they have little or no melanin (Marçon & Maia, 2019). A series of genetic illnesses known as albinism cause little to no melanin synthesis in the body. These disorders are often autosomal recessive. Achromatosis, achromasia, and acromia are other terms for albinism. All races of people are susceptible to this inherited sickness, along with mammals, birds, fish, reptiles, and amphibians. Albinism is a genetic disorder, however in the majority of instances, there isn't always a family history of the condition (Natterson-Horowitz & Bowers, 2020).

To be born with albinism, both parents must have a faulty gene. There is a one in four probability that the child will be born with albinism if neither parent has the condition, but both have the problematic gene. The kind and quantity of melanin produced by a person's body influences their skin, hair, and eye colors. The majorities of albinos are sensitive to sunlight and have a higher risk of skin cancer. A few optical nerves also emerge as a result of melanin. The growth and operation of the eyes are hampered in all kinds of albinism (Bakker et al., 2022).

Albinism is characterized primarily by ocular traits, with or without full symptoms, and is inherited in an autosomal dominant pattern(Neveu et al., 2022). These conditions are not a single illness but rather a group of hereditary ailments with varying manifestations in each individual. The hair and skin of people with ocular albinism may have modest variations from normal to varied pale, whereas those with oculocutaneous albinism (OCA) have a partial or whole lack of pigment in all three body systems(Summers & Adams, 2021). Presently, 15 genes have been related to albinism; however, other genes have been found lately associated to the autosomal recessive OCA with strikingly comparable phenotypic but distinct molecular origins, indicating that there are more than 15 genes which are linked with albinism(De Silva et al., 2021).

Types of Oculocutaneous Albinism

The two types of oculocutaneous albinism are non-syndromic and syndromic, respectively. OCA1, OCA2, OCA3, and OCA4 are four kinds of non-syndromic OCA that are brought on by mutations in the TYRP, TYRP1, and SLC45A2 genes, respectively (Ullah et al., 2022). More phenotypic symptoms, as well as issues with vision and hypo pigmentation, are present in syndromic types of OCA. There are nine distinct Hermansky-Pudlak syndromes (HPS1-9) and Chedaik-Higashi syndromes included in it (CHS). One of the genes in the HPS1 through HPS9 family has mutated, leading to the Hermansky-Pudlak syndrome. These genes all produced proteins that function as endosomal channels (Bowman, Bi‐Karchin, Le, & Marks, 2019; Karampini, Bierings, & Voorberg, 2020). Oculocutaneous albinism in humans is caused by at least 16 genes. Four of them cause non-syndromic oculocutaneous albinism, whereas the other 12 genes cause syndromic oculocutaneous albinism.

Causes of Albinism

Albinism results from a mutation in any one of a number of genes. All of these genes deliver chemically encoded instructions for constructing a different protein that contributes to melanin synthesis. Melanocytes, which may be found in both the skin and the eyes, are responsible for the production of melanin. Depending on the nature of the mutation, it's possible that melanin synthesis might either cease entirely or be drastically reduced (Saud, Sagineedu, Ng, Stanslas, & Lim, 2022).

A person must have two copies of a mutant gene from each parent in order to develop the majority of albinisms (recessive inheritance). A person won't have the condition if they just have one copy (Fischer & Prok, 2020).

The various forms of albinism are caused by distinct genes. The most prevalent kind of albinism is oculocutaneous albinism (OCA). There are other genes that have been identified as potential OCA culprits. Different genes' mutations (changes) result in various forms of OCA. For instance:

• Tyrosinase gene mutations lead to OCA1A and OCA1B.

• The P gene mutations that lead to OCA2

• TRP-1 gene mutations are the cause of OCA3

• SLC45A2 gene mutations result in OCA4 in humans.

OCA is an autosomal recessive disorder; hence it is inherited this way. This implies that in order to have the disorder, a person must inherit two copies of the defective gene—one

from each parent. One in 70 persons are thought to possess the gene that causes OCA.

Those who have the gene are unaffected by the disorder and have normal levels of melanin.

A child's likelihood of having albinism is one in four if both parents contain the gene.

OA - Ocular albinism Two varieties of ocular albinism exist (OA). These are inherited differently and are brought on by various genes:

• OA1 is brought on by a mutation in the GPR143 gene, while AROA is brought on by changes in either the tyrosinase gene or the P gene (Mayo Clinic; NHS UK).

Signs and Symptoms of Albinism

Oculocutaneous albinism is a condition in which a person is born with little to no pigment in their eyes, skin, or hair, or sometimes only their eyes (ocular albinism). Variable levels of pigmentation exist. Some persons age-relatedly develop a little amount of pigmentation in their eyes or hair. On their skin, some people get freckles that are colored. A person who has no melanin at all is often referred to as an "albino." Those having a minimal quantity of melanin are referred to as "albinoid" (Liasis, Handley, & Nischal, 2019)

Albinos are very pale, have fair hair, and have very light eyes. Depending on the level of pigmentation, some people's eyes might seem red or purple. The iris really contains very little color, which explains how this might occur. The blood vessels within the eye may be seen through the iris, giving the eyes a pink or crimson appearance. Albinos are often in comparable health to the general population. However, skin and eye issues are especially prevalent (Crisan & Crisan, 2022).

Skin:

A lighter skin tone is usually the most noticeable indication of albinism; however, this is not always the case. Some people's melanin levels gradually rise over time, darkening their skin tone as they age. A person's skin may burn readily in the sun, which indicates that they are heat sensitive, and their skin seldom tans. Albinism may occur in certain persons after exposure to the sun(Stella).

• Spotty skin (Dark spots on the skin)

• Moles, which are often pink in color owing to the diminished pigment levels.

• Lentigines, which are large patches resembling freckles.

Additionally, skin cancer is more likely to develop. Albinism sufferers should apply sunscreen with an SPF of at least 20 and inform their doctor of any new moles or other skin changes.

Hair:

The color of the hair may vary from white to brown in individuals with albinism. African Americans and Asians often have yellow, brown, or reddish hair. The person's hair color may gradually darken as they age.

Eye color:

Age-related variations in eye color are possible and may range from extremely pale blue, white, or brown.Due to low amounts of melanin in the iris, light reflecting off the retina at the back of the eye may cause the eyes to seem somewhat transparent and appear red or pink under certain lighting conditions.

Since the iris cannot completely screen sunlight due to a lack of pigment, the

individual is sensitive to light. Medics refer to phenomenon as photosensitivity.

Prevalence of Albinisms:

Prevalence of the genes and alleles that cause albinism in a community of Pakistanis.

The relative distribution of albinism gene variations in Pakistani families is shown in (A–C). (A) The distribution of the 94 families examined for the present study's OCA1-4 genes. There are other families listed, along with their percentage contributions, in parenthesis. (B) Our cohort's overall albinism gene pool. (C) Recurrent allele frequencies of the soca gene among Pakistanis (Kildisiute et al., 2021). Results of tetra-primer ARMS tests for recurrent variant identification the positive control applier in all samples produced using the outer primers is represented by the top band in each gel. To create the wild type (Wild) or variant harbouring (Mutant) PCR products, nested allele-specific primers were utilized.

Methodology

The research used a cross-sectional design to examine the prevalence of albino patients in the district of KhairPur. Only albinos were included in the study's sample. Data from the district of KhairPur was gathered to determine the frequency of albino patients. Data was gathered between July 2022 and Male and female patients who had been treated outdoors were regarded as being of a different age. A total of samples were collected during the research period, and a set of questionnaires based on the names, addresses, ages, levels of education, genders, and family histories of albanic patients were constructed. Nevertheless, the district of Khairpur’s albino patients' predominance of albinism records was gathered.

In the Sukkur division of the Pakistani province of Sindh, there is a district known as Khairpur. It is situated halfway between Sindh's central and northern sides. It is bordered on the north by the districts of Shikarpur and Sukkur as well as by India; on the south by the districts of Singhar and Nawab Shah; and on the east by the districts of Larkana, Naushero Feroz, and the Indus River. This district's updated area is 15910km2. Its latitudes are around 27.529951 and 68.758141. Khairpur district has a population of 2405190 people as of the 2017 census, with 1240254 men and 1164826 women. Due to Mir (talpurs) conquest, the Khairpur district is also known as KhairPur Mir's.

Results

Finding out the knowledge and attitudes concerning albinism disorders and the prevalence of albinos in the district of Khairpur is the purpose of our study.

According to test findings, the majority of albinism patients in the city are between the ages of 6 and 15, with a high valid percentage of 75% and a frequency of 15 out of 20. Table 1 indicates the ratio of various age groups of albinism patients. The cumulative frequency as a whole is 100.0 percent.

Age	Frequency	Percent	Valid percent	Cumulative percent
5-6 days	1	5%	5%	5%
6-15 years	15	75%	75%	80%
16-34 years	4	20%	20%	100%
Total	20	100%	100%	

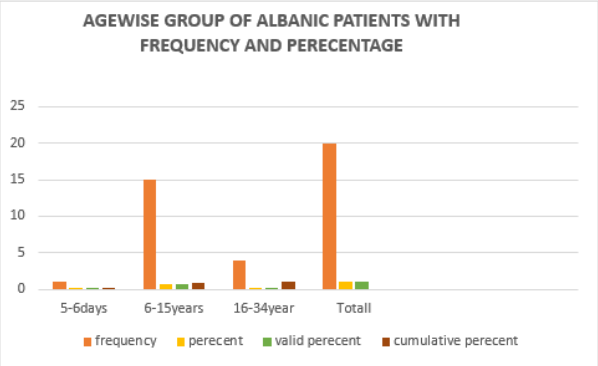
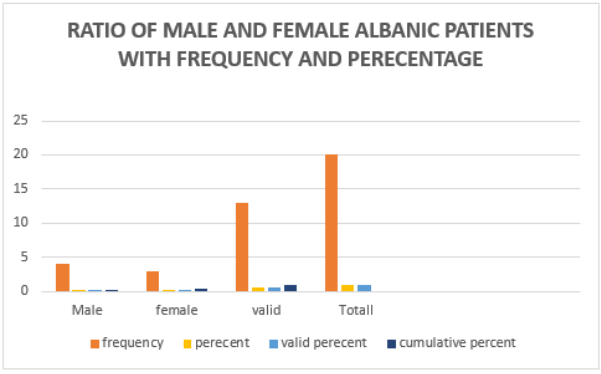


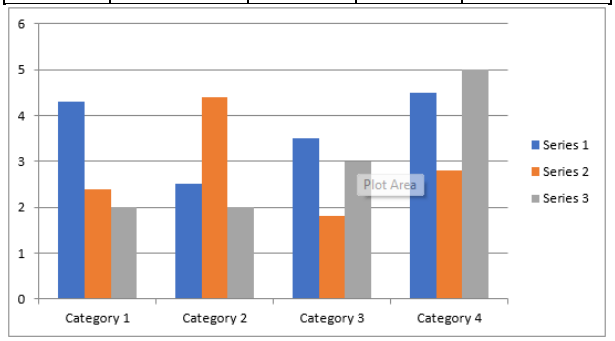
Table 2 shows the proportion of albinism patients by gender. With a valid proportion of 20%, the majority of patients are male, while the female ratio is 15%. In the city, 65 percent of children are afflicted with this condition. 100 percent is the whole percentage.

Gender	Frequency	Percent	Valid percent	Cumulative percent
Male	4	20%	20%	20%
Female	3	15%	15%	35%
Valid	13	65%	65%	100%
Total	20	100%	100%	



The prevalence of albinism sickness is broken down by marital status in Table 3. Twenty people, or 7 of them, have this illness; the actual prevalence is 35%. While there are 1 marriage per 100 people with a legitimate proportion of 5%. According to the findings, children suffer the most—12 times out of every 100—with a valid proportion of 60%. 100 percent is the total percentage.

Status	Frequency	Percent	Valid percent	Cumulative percent
Single	7	35%	35%	35%
Married	1	5%	5%	40%
Child	12	60%	60%	100%
Total	20	100%	100%	



The test results for patients with albinism's family history are shown in Table 4. According to the test findings, the frequency of the illness in the mother and father is one, with a valid percentage of five percent, while the frequency in the brother and sister is high, at 18, with a valid percentage of ninety percent. As a whole, though, the proportion is 100%.

Family	Frequency	Percent	Valid percent	Cumulative percent
Mother	1	5%	5%	5%
Father	1	5%	5%	10%
Brother & sister	18	90%	90%	100%
Total	20	100%	100%	

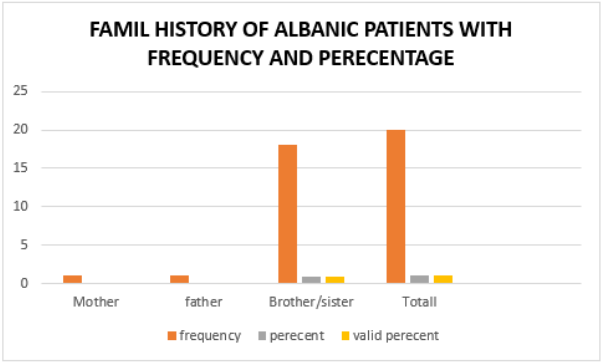


Table 5 shows the percentage of albinism subtypes in the family. The findings show that Type 2 with a frequency of 9 and a valid percentage of 45% is to blame for the majority of family distress. While the type 1 has a frequency of 8 and a validity rate of 40%. The cumulative percentage is 100%.

Type of albinism patients in family	Frequency	Percentage	Valid percent	Cumulative percent
Type 1	8	40%	40%	40%
Type 2	9	45%	45%	85%
Valid	3	15%	15%	100%
Total	20	100%	100%	

Discussion

The goal of our study is to determine how people feel and know about albinism, its disorders, and how common it is in the Khairpur district. White or brown hair, feathers, scales, skin, and pink, brown, or blue eyes are all signs of albinism, which is the amiable lack of melanin (the skin pigment) in an animal, a plant, or a human.

Our findings show that most of the patients are belongs to the age group of 6-15 with the percentage of 75, Which is highest in our findings. As per findings most of the patients are male excluding children male has the percentage of 20% while the female has 15%, in comparison children have high percentage i.e., 65%.

According to test findings for the patients' marital status, the majority of patients—35 percent—are single. While the married enjoy a 5 percent advantage. Children has a high proportion, 60 percent, indicating that the majority of patients are suffering from this disease while they are younger. As per the outcomes of the family history, the majority of patients—90%—are brothers and sisters. The proportion of type 2 patients is high, at 45%.

Albinism (albino) is genetic disorder diseases that affect the human and animal body

It is possible to regulate the genetic transmission of albinism. Due to a lack of pigment, most albinism patients exhibit more symptoms (brown hair, a white body, and green eyes). Albinism is passed on from parents to children. The Alb flora is included in the study's findings (Hong et al., 2018).

Due to a total lack of pigment, individuals with Ocular 0A and Oculocutaneous albinism (OCA) will have white skin, white hair, and light eyes (webM And LLC 2019). Another topic that was explored in this research was the distribution of instances of albinism. Secondary data reveal that district Khairpur had 40% of the albino cases and Talka Kotdeji had 60% of the cases.

Conclusion

It was concluded from this study that the participants had high knowledge and a positive attitude about albinism diseases. In secondary data, the proportion of pediatric patients was higher than that of male and female patients. Patients with albinism participated in surveys at higher rates and provided responses reflecting their attitude and degree of knowledge. According to secondary statistics, the proportion of patients who were children at their highest was 65%, men at their greatest was 20%, and women at their lowest proportion was (15 percent). The patient age comparison reveals that patients with recorded instances of albinism were under 30 years old. With a ratio of (60%) the most contaminated instances of albino patients were in the tahsil kotdeji District Khairpur.

Recommendations

Since illiteracy is the driving factor behind beliefs and attitudes concerning albinism, illiteracy must be eradicated. A strategy for a long-term solution should be developed to ensure that students in elementary schools are aware of albinism. This will significantly improve the knowledge of the albinism condition among those with a low level of

education. Awareness of albinism should be supported by the socialization of youngsters with good attitudes regarding the condition. This will assure a society with good views about albinism and other types of disability. This will broaden the awareness of albinism and assist young people in rejecting cultural misconceptions about the condition's cause.

Informed Consent Statement:

All participants in the research gave their permission after being fully informed.

Acknowledgments:

We are thankful of the participants' willingness to cooperate.

Conflicts of Interest:

The authors state that they have no conflicts of interest.

Reference

Bakker, R., Wagstaff, E.L., Kruijt, C.C., Emri, E., van Karnebeek, C.D., Hoffmann, M.B., Brooks, B.P., Boon, C.J., Montoliu, L., van Genderen, M.M. and Bergen, A.A., 2022. The retinal pigmentation pathway in human albinism: Not so black and white. Progress in Retinal and Eye Research, 91, p.101091.Bastonini, E., Bellei, B., Filoni, A., Kovacs, D., Iacovelli, P., & Picardo, M. (2019). Involvement of non‐melanocytic skin cells in vitiligo. Experimental Dermatology, 28(6), 667-673. https://doi.org/10.1016/j.preteyeres.2022.101091

Bowman, S.L., Bi‐Karchin, J., Le, L. and Marks, M.S., 2019. The road to lysosome‐related organelles: Insights from Hermansky‐Pudlak syndrome and other rare diseases. Traffic, 20(6), pp.404-435.Crisan, D., & Crisan, M. (2022). Dermatologic Concepts and Terminology. In Textbook of Dermatologic Ultrasound (pp. 21-72). Cham: Springer International Publishing. https://doi.org/10.1111/tra.12646

De Silva, S.R., Arno, G., Robson, A.G., Fakin, A., Pontikos, N., Mohamed, M.D., Bird, A.C., Moore, A.T., Michaelides, M., Webster, A.R. and Mahroo, O.A., 2021. The X-linked retinopathies: physiological insights, pathogenic mechanisms, phenotypic features and novel therapies. Progress in Retinal and Eye Research, 82, p.100898.Fischer, A., & Prok, L. D. (2020). DISORDERS OF PIGMENTATION. Dermatology Secrets E-Book.

Hong, E.S., Zeeb, H. and Repacholi, M.H., 2006. Albinism in Africa as a public health issue. BMC public health, 6(1), pp.1-7.Karampini, E., Bierings, R., & Voorberg, J. (2020). Orchestration of primary hemostasis by platelet and endothelial lysosome-related organelles. Arteriosclerosis, Thrombosis, and Vascular Biology, 40(6), 1441-1453.

Kildisiute, G., Kholosy, W.M., Young, M.D., Roberts, K., Elmentaite, R., van Hooff, S.R., Pacyna, C.N., Khabirova, E., Piapi, A., Thevanesan, C. and Bugallo-Blanco, E., 2021. Tumor to normal single-cell mRNA comparisons reveal a pan-neuroblastoma cancer cell. Science advances, 7(6), p.eabd3311.Liasis, A., Handley, S. E., & Nischal, K. K. (2019). Occipital petalia and albinism: a study of interhemispheric VEP asymmetries in albinism with no nystagmus. Journal of Clinical Medicine, 8(6), 802. 10.1126/sciadv.abd3311

Marçon, C.R. and Maia, M., 2019. Albinism: epidemiology, genetics, cutaneous characterization, psychosocial factors. Anais brasileiros de dermatologia, 94,

pp.503-520.Natterson-Horowitz, B., & Bowers, K. (2020). Wildhood: the astounding connections between human and animal adolescents. Scribner. https://doi.org/10.1016/j.abd.2019.09.023

Neveu, M.M., Padhy, S.K., Ramamurthy, S., Takkar, B., Jalali, S., Cp, D., Padhi, T.R. and Robson, A.G., 2022. Ophthalmological manifestations of oculocutaneous and ocular albinism: Current perspectives. Clinical Ophthalmology, pp.1569-1587.Saud, A., Sagineedu, S. R., Ng, H. S., Stanslas, J., & Lim, J. C. W. (2022). Melanoma metastasis: What role does melanin play?. Oncology Reports, 48(6), 1-10.

Stella, A.E., 2021. Biological and Social Challenges of the Aging Skin in Older Africans. Asian Journal of Research in Dermatological Science, 4(2), pp.7-20.Summers, C. G., & Adams, D. R. (2021). ALBINISM: OCULAR AND OCULOCUTANEOUS ALBINISM AND HERMANSKY–PUDLAK SYNDROME. Cassidy and Allanson's Management of Genetic Syndromes, 45-59. http://journal.librarykeep.com/id/eprint/209

Ullah, M.I., 2022. Clinical and Mutation Spectrum of Autosomal Recessive Non-Syndromic Oculocutaneous Albinism (nsOCA) in Pakistan: A Review. Genes, 13(6), p.1072. https://doi.org/10.3390/genes13061072