Oculocutaneous Albinism

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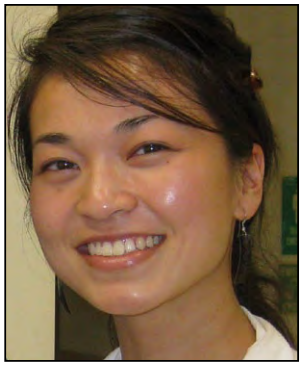

As residents, we can all relate to the eureka moment when a patient with a condition we have studied but have never seen in practice is standing in front of us. All of the memorized snippets, word associations, and Kodachrome images piece together. The condition's sequelae and effects on quality of life also become clearer. I had this experience understanding albinism as a medical student following a rotation in Lilongwe, Malawi. The rotation, which was organized by Aisha Sethi, MD, from the University of Chicago, Illinois, was superb on multiple levels including the broad range of dermatologic conditions, the challenge of treating and diagnosing patients despite limited resources, the lifelong friendships that were formed, and the opportunity to meet frontline dermatologists in Lilongwe and see an inspiring albinism awareness initiative in action.

Overview of Oculocutaneous Albinism

Oculocutaneous albinism (OCA) is a genetic pigmentation disorder resulting in diffuse hypomelanosis. Patients with albinism have normal numbers of epidermal and follicular melanocytes, but melanin pigment is totally or partially absent.[1] The 4 main types of OCA are autosomal recessive (Table). Diagnosis can be made based on the presence of typical cutaneous and ocular findings. Distinguishing between subtypes based on clinical features can be difficult due to overlap in clinical phenotypes.[3,4] The extent of morbidity is multifactorial, influenced by phenotype, education, and resource availability.

The worldwide incidence of albinism is 1 in 20,000.[1] The incidence is lower in the United States (1 in 37,000[5]) and higher in sub-Saharan Africa (Zimbabwe: 1 in 1000 to 1 in 4000; Tanzania: 1 in 1400[6]), which could be due to consanguinity.[7] The incidence of OCA types varies by population. Oculocutaneous albinism type 2 is the most common form of OCA worldwide in Africans and African Americans.[3,8,9] In Japan and China, OCA types 1 and 4 have been found to be the first and second most prevalent forms of OCA, respectively,[10-12] though type 4 usually is considered rare worldwide.[11] A US study found OCA type 1 to be most prevalent,[13] while a Pakistani population study found OCA types 1 and 2 to be most common.[14]

Clinical Implications of Albinism

Due to reduced or absent melanin, albinos are highly susceptible to the harmful effects of UV radiation and are at a higher risk for actinic injury (eg, sunburn, blisters, lentigines, solar elastosis, basal cell carcinoma, squamous cell carcinoma).[6,15-17] Many albinos in African populations develop actinic keratoses or malignancies by 20 years of age.[18,19] Studies found squamous cell carcinoma to be the most common form of skin cancer among Africans with albinism, most often on the head and neck. Sequelae from skin cancer is a major cause of early death in patients with albinism.[16,17] It is crucial to educate albinos and their family members about the importance of sun avoidance, barrier protection, and sunscreen use. Unfortunately, many sun protection products (eg, sunscreen, wide-brimmed hats) can be expensive.

Melanin plays an important role in the development of the optic system. Patients with albinism experience light sensitivity, refractive errors, and reduced visual acuity due to foveal hypoplasia and nystagmus. The misrouting of optic nerve fibers at the chiasm results in nystagmus and strabismus.[1,20] When possible, schools should be informed that a child with albinism may have low vision. Patients may benefit from low-vision aids such as glasses for long or short

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Types of Oculocutaneous Albinism[1,2]

Type	Gene	Clinical Phenotype
OCA type 1A	*TYR*	Absent tyrosinase activity; white hair and skin, blue eyes, and amelanotic nevi; most severe visual defects
OCA type 1B	*TYR*	Reduced tyrosinase activity; no pigment at birth (similar to OCA type 1A); may develop some pigment, ephelides, and melanocytic nevi throughout life; can tan; visual defects not as severe
		OCA 1B subtype (temperature-sensitive OCA): tyrosinase activity >35°C, thus extremities and head may develop pigment later in life
OCA type 2	*OCA2* (formerly called *P* gene)	Abnormal processing and transport of tyrosinase; broad phenotype (no pigment to normal pigment); can have nevi and ephelides
OCA type 3	*TYRP1*	Delayed maturation and early degradation of tyrosine; reddish brown skin referred to as rufous or red OCA; vision may be normal
OCA type 4	*SCL45A2* (also called *MATP*)	Defects in transporter protein; variable hypopigmentation; phenotypically similar to OCA type 2

Abbreviations: OCA, oculocutaneous albinism; *TYR*, tyrosinase; *OCA2*, oculocutaneous albinism II; *TYRP1*, tyrosinase-related protein 1; *SCL45A2*, solute family carrier 45, member 2; *MATP*, membrane-associated transporter protein.

distances and/or tools to enhance contrast between black, white, and colors. There also are computer programs available that can magnify and/or verbalize text.

One of the most important messages to relay to patients with albinism and their family members is that they are not abnormal. Although the most common cause of early death in albinos is progressive skin cancer, patients can have normal life spans with proper skin care. They also develop normally and have normal intelligence; appropriate visual aids ensure students the best chance of keeping up with peers in school. They also have normal fertility.[4]

Albinism in Sub-Saharan Africa

Regardless of location, albinism can leave patients feeling stigmatized and isolated. Thus albino awareness initiatives are important everywhere and are particularly meaningful in sub-Saharan Africa. The disease can take a toll on health and education; for instance, teachers may not recognize a student's low vision or a family might lack the tools to help the child, making it difficult to succeed in school. Already facing prejudice and discrimination in the workplace, minimally educated individuals with albinism often take jobs that involve being outdoors, including roadside trading, market stalls, and agricultural day labor. Patients who are not employed are still exposed to the sun, surviving by farming small plots.[21] Folklore and pervasive beliefs indicate that albinos have magical powers. Some men believe having sex with an albinistic woman can cure illnesses such as human immunodeficiency virus.[22] Numerous albinos in Tanzania and Burundi have been murdered at the hands of hunters who can earn as much as $75,000 for body parts used as good luck charms.[7] A 2009 report by the International Federation of Red Cross and Red Crescent Societies noted that an undetermined number of Tanzanian albinos had been displaced and were clustered near police stations and churches, unable to roam freely out of fear for their own safety.[21]

Many organizations in addition to the Red Cross and media are working hard to correct the situation. In 2007, Dr. Sethi founded the albinism clinic in

Kamuzu Central Hospital in Lilongwe and the first annual Albinism Awareness Day. With the help of committed dermatology officers (ie, Malawian providers who received dermatology training at the Regional Dermatology Training Centre at the Kilimanjaro Christian Medical Centre in Moshi, Tanzania), the hospital continues to hold year-round weekly clinics for albinistic patients and to recognize Albino Awareness Day annually, ensuring that patients can socialize and receive free skin examinations and sun protection gear.

Another important program is Hats On For Skin Health (http://www.hatsonforskinhealth.org), a global campaign that uses donations to purchase hats and sun-protective items to be distributed by the Regional Dermatology Training Centre in Tanzania. The albino assistance program includes a mobile skin care clinic that visits surrounding villages, providing skin examinations, supplies, and education.[7,23] Additionally, Asante Mariamu (http://www.asantemariamu.org), which was founded by a mother of 2 children with albinism living in Virginia, is a grassroots organization dedicated to raising awareness about the immediate and long-term threats to albinos living in East Africa.

Resources for Patients

In addition to counseling our albinistic patients about their condition, dermatologists also can play an important role in connecting patients with support groups and resources. A prominent support group in the United States is the National Organization for Albinism and Hypopigmentation (http://www.albinism.org), which has a comprehensive Web site that provides educational information on albinism with links to other resources. This organization also organizes conferences, teleconferences, webinar series, family summer camps, and adult weekend excursions, and also offers scholarships to students with albinism. The Vision for Tomorrow Foundation (http://www.visionfortomorrow.org) is a US organization dedicated to vision problems related to albinism. The Albinism Fellowship (http://www.albinism.org.uk) provides information, advice, and support for albinos and their family members.

You also can encourage your patients to visit the Positive Exposure Web site (http://www.positiveexposure.org), which was founded by former fashion photographer Rick Guidotti. The organization has made huge strides in showing the world that albinism and beauty are not mutually exclusive. The program utilizes photography and video to change public perception of people living with genetic, physical, and behavioral conditions, including albinism. Patients with albinism also may be interested in learning more about the many world famous and successful individuals with albinism, such as fashion models Connie Chiu, Diandra Forrest, and Shaun Ross; comedian Victor Varnado; musicians Edgar and Johnny Winter; and Tanzanian politician Salum Khalfani Bar'wani.

Conclusion

We are bound to care for patients with albinism in our home countries. Since my trip to Malawi, I have had 1 patient with Hermansky-Pudlak syndrome and 2 patients with OCA (one at a free clinic and the other in an insured office setting). All 3 patients had some form of actinic injury and/or malignancy, though not nearly as much damage as the patients we saw in Malawi, most likely because of access to better resources in the United States. Because of my experience in Malawi, I was able to quickly counsel the patients and direct them to several resources.

With any genetic condition, a patient's quality of life can be remarkably improved with proper support and education. For me, one of the most important reminders for patients is that there is an incredibly strong and dedicated support network for albinos that goes beyond the organizations and resources that are discussed in this article. I invite you to explore others and share this knowledge with patients of your own!

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