International Journal of Multidisciplinary Approach and Studies

ISSN NO:: 2348 – 537X

Know more about ALBINISM

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ABSTRACT

Albinism is one of the rarest genetic disorders. It is independent of the population, religion and type and standard of living. Albino individuals, be it human or animals are less fit and unhealthy. Talking about their appearance, albino individuals have yellowish to pale skin , white or very lightly tinted hair and are mostly born with no pigment in their eyes, giving them a "Red Eye" look. Being a genetic disorder, it's a recessive allele character which means an individual albino can be born to a non albino parent but both the parent must have been a carrier to the defective gene. Albino individuals are more susceptible to diseases and most commonly eyes and skin diseases like scattering of light in eyes, sunburn and freckles on the skin. Some albino individuals also develop a little pigment with age but such individuals are still considered under albino cases.

INTRODUCTION

Albinism also known as achromasia, achromia or achromatosis is a congenital disorder which is signalized by the complete or partial absence of pigment in the hair, skin and eyes due to absence of tyrosinase, a copper-containing enzyme that is involved in production of melanin. Albinism is also caused from inheritance of recessive gene alleles which is known to affect all vertebrates as well as humans. While an organism with complete absence of melanin is called an albino an organism with only a diminished amount of melanin is described as albinoid [*Tietz, W. A Syndrome of Deaf-Mutism Associated with Albinism Showing Dominant Autosomal Inheritance. Department of Pediatrics, Southern California]*. Albinism is kindred with many vision defects like, photophobia, astigmatism and nystagmus. Lack of skin pigments makes people suffering more susceptibility to sunburn and skin cancers. Though a hereditary condition, in most of the cases, there's no history of albinism in the family.

PRENATAL TESTING

For couples with no child suffering from albinism, there is no effortless test to govern whether the fetus carries a defective gene for albinism.In the case of parents who already have a child with albinism, It is feasible to test using either amniocentesis or chorionic villous sampling (CVS). Cells in the fluid are scrutinized to see if they have an albinism gene from each parent.

CAUSES

A person inherits the defective genes that cause him incompetent to produce the normal amounts of melanin. Various genes are involved in albinism, based on its specific type.

Genes carry the information that makes you an individual. We have two copies of these autosomal chromosomes and genes: one inherited from our father, and the other from our mother. Albinism is a "recessive trait" which means a person without being affected by albinism can carry its trait. Both mother and father must carry a defective gene to have a child with albinism. When none of the parent has albinism but they both carry a defective gene, there is a one in four chance that the baby will be affected with albinism.

SIGNS AND SYMPTOMS

Since nativity, infants with albinism have negligible pigmentation in their eyes, hair and skin (oculocutaneous albinism) or sometimes only in the eyes (ocular albinism).Most people with oculocutaenous albinism appear totally white cause the melanin pigments accountable for black, brown or some blonde colors are absent. Ocular albinism out-turns in pale blue eyes which may entail genetic testing to diagnose. As the individuals with albinism have skin that completely lacks melanin, that helps to protect the skin from the sun's ultraviolet radiations, their skin can burn more easily from overexposure to sun. The amount and intensity of pigmentation varies. Some people secure minor pigmentation in their hair or eyes with age. Some people develop pigmented freckles on skin. Albino people are very pale with average hair color and very light colored eyes. In some, eyes appear purple or red, depending upon the amount of pigmentation. This happens because the iris actually has very less color therefore, the eyes appear red because the blood vessels inside from the eyes show through the iris.

A person with albinism is altogether as fit and fine as the rest of the population. However, problems with eyesight and skin are exceptionally common. Lack of pigment in the eyes also upshots in problems with vision. Those affected with albinism are generally as healthy as the rest of the community with growth and development arising as normal and albinism itself do not cause mortality.

VISUAL PROBLEMS

Development of optical system is greatly reliant on the presence of melanin and the cut back or lack of this pigment in albino individuals may generate an eye conditions common in albinism. Some of the eyesight problems related with albinism emerge from a poorly developed Retinal Pigment Epithelium (RPE) due to the lack of melanin. This degenerate RPE causes foveal hypoplasia (a failure in the development of normal foveae) which results in eccentric fixation and lowers the visual activity.

In albino individuals, the iris lack considerable pigment to block the light, therefore causing the decrease in pupil diameter, reducing the amount of light entering the eye. Furthermore, the poor development of the RPE, which in normal eyes absorbs most of the reflected sunlight, further increases flare due to light scattering within the eye. The consequent sensitivity (photophobia) generally leads to discomfort in bright light, but this can be reduced with the use of sunglasses.

GENETICS

Occulocutaenous albinism is in majority cases is the result of the biological inheritance of genetically recessive alleles inherited from both the parents of an offspring, though some rare forms are inherited from only one parent. There are also other genetic mutations which are found to be integrated with albinism.

The possibility of individual with albinism resulting from the pairing of an organism affected with albinism and the one without albinism is very less. Nevertheless, because organisms can be carriers of genes for albinism without showing any sign, albino offspring can be produced by two non-albino parents. Albinism normally shows with equal frequency in both sexes. An exception to this is ocular albinism that is inherited by an offspring through X-linked inheritance. Thus, ocular albinism is seen more frequently in males as they have a single X and Y chromosome, unlike females, who have two X chromosomes.

There are two different types of albinism: a partial lack of the melanin is known as hypomelanism, or hypomelanosis and the total absence of melanin is known as amelanism or amelanosis.

DIAGNOSIS

Genetic testing can confirm albinism and what variety it is. The symptoms of albinism can be treated by a few methods given below.

TREATMENT

Treatment of the eye conditions basically comprises of visual rehabilitation. Surgery is possible on the ocular muscles, to reduce the "shaking" of the eyes back and forth in case of nystagmus. The potency of all these procedures varies greatly and resting on individual circumstances.

Glasses and other vision aids, large-print materials as well as bright but angled reading lights, can help individuals with albinism, even though their vision cannot be corrected completely. Some people with albinism do work with reading glasses, or hand-held devices such as magnifiers or colored contact lenses to block light transmission through the iris. But this remedy is not possible in case of nystagmus, due to the irritation that is caused by the movement of the eyes.

EPIDEMIOLOGY

Albinism influence people of all ethnic frameworks, its frequency worldwide are evaluated to be approximately 1 in 17,000. Widespread presence of the different forms of albinism varies considerably by population and is the highest overall in people of sub-Saharan African descent.

TYPES OF ALBINISM

The system for classifying types of albinism is based basically on which mutated gene caused the disorder rather than by exterior signs. However, most types of albinism have some specific features that can differentiate them from one another. Types of albinism incorporate:

- **Oculocutaneous Albinism-** Oculocutaneous albinism is caused by a mutation in one of four genes. These mutations result in character and symptoms related to vision (ocular) and those related to skin, hair and iris color.

- **X-linked ocular albinism-** The cause of X-linked ocular albinism, which occurs almost extensively in males, is a gene mutation on the X chromosome. People who have ocular albinism have the developmental and functional eyesight problems. But hair,skin and eye color are generally in the normal or a bit lighter than that of others in their family.
- **Hermansky-Pudlak syndrome-** Hermansky-Pudlak syndrome is a rare albinism disorder caused by a mutation in one of eight genes corresponding with this syndrome. People with this disorder have symptoms like the one with oculocutaneous albinism, but they also develop lung ,bowel diseases and a bleeding disorder.
- **Chediak-Higashi syndrome-** Chediak-Higashi syndrome is a rare form of albinism that's associated with a mutation in the LYST gene. Signs are similar to those of oculocutaneous albinism. The hair looks like brown or blond with a silvery tint, and the skin is mostly creamy white to grayish. People with this syndrome have a defected white blood cells(WBCs) that results in a susceptibility to infections.

GUINNESS WORLD RECORD CASE REPORTED IN INDIA

A white-skinned Indian couple are set to enter the record books along with their offspring, after becoming the world's biggest albino family.
The ten members of the Pullan family, headed by Rosetauri, 50, and his wife Mani, 45, all have the extremely pale skin and near-white hair of albinos.
But despite years of prejudice and suffering the poor vision which is a side effect of the condition, the Pullans and their eight other family members are set to land a Guinness World Record. The Pullan's sons Shankar, 24, Vijay, 25 and Ramkishan, 19, and daughters Renu, 23, Deepa, 21 and Pooja, 18, inherited their albinism, according to The Sun.
Renu married a man who also had albinism, Rosheh, 27, and the couple's son Dharamraj, 2, also inherited the condition from his parents.

EPIDEMIOLOGIC DATA ON ALBINISM

Epidemiologic data on albinism from a public survey in African countries,published by the World Health Organization (WHO) in 2006, mentions that: “Epidemiologic data on albinism, such as prevalence, were available for South Africa, Zimbabwe, Tanzania and Nigeria. Prevalence as high as 1 in 1,000 were reported for selected populations in Zimbabwe and other specific ethnic groups in Southern Africa. An overall estimate of albinism prevalence ranges from 1/5,000 – 1/15,000”.[1] According to WHO, the estimated prevalence of albinism suggests the existence of tens of thousands of people living with albinism in Africa. OHCHR did not receive detailed data on the prevalence of albinism from any country or region.

REFERENCES:

i. "Albinism—Review of Optometry Online".

ii. King RA, Hearing VJ, Creel DJ, Oetting WS: Albinism, 4353-4393, in Scribner, Charles R et al, The Metabolic and Molecular Bases of Inherited Disease, 7th ed., McGraw Hill, Inc., Health Professions Division, New York 1995.

iii. Albinism: MedlinePlus Medical Encyclopedia. (n.d.). National Library of Medicine - National Institutes of Health. Retrieved April 5, 2012, fromhttp://www.nlm.nih.gov/medlineplus/ency/article/001479.htm

iv. "Albino" Dictionary.com. Accessed May 11, 2011

v. “Pronunciation of albino" Macmillan Dictionary. Accessed May 11, 2011

vi. http://www.dailymail.co.uk/news/article-2111298/Meet-Indian-couple-children-set-new-record--worlds-biggest-ALBINO-family.html

vii. http://www.medicalnewstoday.com/articles/245861.php