Research Paper

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A Black and White Twin

Medical Science

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ABSTRACT

Albinism is an autosomal recessive disorder that is caused by a defective synthesis of melanin, resulting in a generalized reduction of pigmentation in the skin, hair and eyes, and leading to an increased risk of skin cancer and vision problems. We report a case of a 26-year-old multigravida who delivered diamniotic twins: two sons were born with a totally different appearance. The first child had a light brown skin, black hair and black eyes, whereas the second had a striking white skin and eyes. Oculocutaneous albinism (OCA) and heteropaternal superfecundation were considered in the differential diagnosis. Genetic testing confirmed the diagnosis of OCA type 2 in the second child. The diagnosis of albinism has clinical implications and must be considered when a black and white twin is born.

INTRODUCTION:

Albinism is an autosomal recessive disorder that is caused by defective synthesis of melanin, resulting in generalized reduction of pigmentation in skin, hair, eyes and leading to an increased risk of skin cancer and vision problems. 1 .it is genetically determined heterozygous group of disorder involving deficiency in enzyme tyrosinase, which mediated conversion of tyrosine to melanin. The two main tpes are: oculocutaneous and ocular. Oculocutaneous can either be tyrosinase negative or tyrosinase positive.2

Ocular albinism: the pattern of inheritance in ocular albinism is either X linked or autosomal recessive. The skin and hair in ocular albinism appear normal but still typical signs and symptoms present

Oculocutaneous albinism: the hereditary pattern of albinism is usually autosomal recessive. The tyrosinase negative and tyrosinase positive form represent genotypes.

The tyrosinase negative type is also called complete albinism. These individuals are uncapable to synthesize melanin. The tyrosinase positive type is called incomplete albinism. Tyrosinase positive albinos can synthesize variable amount of melanin and are therefore harder to diagnose.

Tyrosinase negative: Have straw or platinum blond hair and pale skin. Tyrosinase negative burn in sun and never tan. They have typically blue to pink appearing iris which transilluminate completely.

Tyrosinase positive: Has also light hair and eye color. But the color of hair can darken with age.

In both types, ocular findings include: reduced central vision, nystagmus, iris transilumination, decreased pigmentation of fundus, poor foveal development and ill defined macular landarks.3

Albinism affects people from all races and it is estimated that the incidence of albinism in the general population is approximately 1:17,000 4, 5this incidence appears lowest amongst Asians 5

CASE REPORT:

A 26 year old healthy multi-gravida of Indian origin, spontaneously conceived diamniotic twins. Pregnancy was uneventful. Induction of labor followed with a subsequent caesarean section and two sons were born with Apgar scores at 1 and 5 min of 8 and 9 and 10 and 10 respectively. One of the twins at the age of one month was bought to the hospital with septicemia. At physical examination, great difference between child I and child II were noticed. Child I had brown skin with black hair and black eyes, whereas child II had a striking white skin and eyes

An explanation for the great difference in appearance between the two children could be genetic, as on taking medical history the father revealed having family history of albinism on paternal side with himself being an albino. Ophthalmologic examination of the albino child (child II) at the age of 1 month confirmed a blue iris; no pigment was observed in the fundus and the papilla was grayish. These findings correspond to the clinical picture of oculocutaneous albinism (OCA).

DISCUSSION:

Albinism affects people of all ethnic backgrounds; its frequency worldwide is estimated to be approximately one in 17,000. Prevalence of the different forms of albinism varies considerably by population, and is highest overall in people of sub-Saharan African descent6. It is an autosomal recessive disorder that is caused by a defective synthesis of melanin. In the synthesis of melanin, enzyme tyrosinase is of major importance, because it conducts the first step of the melanin biosynthetic pathway. There are different types of OCA. Most relevant are OCA types 1 and 2. Type 1 is called the tyrosinase negative form, and is divided into 1A and 1B, and these two types result from a mutation of the tyrosinase gene (TYR gene) on chromosome 11. In type 1A, the TYR gene produces a completely inactive tyrosinase enzyme, and hence no melanin is formed in any melanocyte. In type 1B, the TYR gene produces a partially active tyrosinase enzyme, resulting in a diminished synthesis of melanin. In OCA type 2, a mutation in the OCA 2 or P gene on chromosome 15 is present. The OCA 2 protein produced is important for normal biogenesis of melano- somes, and for normal processing and transport of melanosomal proteins such as tyrosinase. Owing to the OCA 2 mutation, tyrosinase is retained in perinuclear compartments, resulting in a diminished synthesis of melanin. As functional tyrosinase is produced in OCA type 2, it is named the tyrosinase-positive form.7,8OCA types 1 and 2 differ on the bases of genetics and the degree of skin and hair hypopigmentation and ocular manifestations, with OCA type 1 being more severe. Genetic testing, by demonstration of the mutated TYR or OCA 2 gene in a blood sample, confirms the diagnosis of albinism. Prenatal diagnosis of albinism is also possible by demonstration of the mutated tyrosinase gene after amniocentesis or from a fetal skin biopsy.7However, albinism is merely clinically diagnosed by the patient's appearance, being characterized by a general reduction of pigment in the skin, hair and eyes. Exposure of the non- or reduced pigmented skin to sun radiation leads to sunburns, pigmented spots and skin cancer. Ocular manifestations of albinism include photophobia with painful eyes because of hypopigmentation and translucency of the iris. The retina is damaged by defective filtration of ultraviolet radia-

tion by the iris, leading to decreased visual acuity (usually in the range 20/60 to 20/400) or even blindness. The vision problems are mainly caused by foveal hypoplasia and misrouting of the optic nerve fibers, because of the absence or reduced presence of melanin. Foveal hypoplasia leads to an inaccurate projection of the image on the macula lutea because of the widespread localization of the cones, resulting in reduced vision and nystagmus that presents from the age of 3 to 4 months.7,8 There is no curative treatment for albinism. Photoprotection, however, can reduce the main complications such as skin cancer and loss of vision. Examples of preventive measures are covering clothing, sunglasses and sunscreen. An annual check-up of the skin is advised for early diagnosis and treatment of premalignant or malignant skin disorders. Improvement of the vision can be accomplished by means of sunglasses, the use of indirect light and contrast, and of course optimal dioptry correction. Complaints of nystagmus can be treated with contacts or surgical correction of the eye muscles.7,8 The most prevalent skin disorders in albinos are basal cell carcinoma and squamous cell carcinoma, whereas melanoma are rare.7The highest prevalence of squamous cell carcinoma is consistently shown in the albino population.9–12Lookingbill et al.11 studied albinos in Tanzania and found actinic keratosis in 91% of the albino patients of 20 years and older, and in 34% of the patients who were 30 years and older they diagnosed skin cancer. Death from skin cancer resulted from erosion through the eyes, skull or large blood vessels. This occurred with squamous cell carcinoma, whereas the basal cell carcinoma is less aggressive and does not metastasize.7Aquaron studied 273 patients with OCA with a 15-year follow-up, and concluded that the majority of the albinos die from skin cancer.11Albinism is a rare disease, but the increased risk of skin cancer and vision problems indicates the importance of the diagnosis. When a black and white twin is born, one should consider the diagnosis albinism.

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