Introduction

Oculocutaneous albinism (OCA) is part of a heterogeneous group of genetic conditions which occur in autosomal recessive manner. Three main types of OCA have been identified: tyrosinase-negative (OCA1), also known as BOCA (“brown OCA”); tyrosinase-positive (OCA2); and rufous OCA, also called ROCA.

In all these cases there is an alteration of the production of melanin pigment in the biosynthetic process in which the enzyme tyrosinase is involved.

In the form OCA1 there is an almost total absence of melanin or only a minimum quantity. It is a rare form but, from a phenotypic point of view, the most striking. Affected individuals have pale straw-coloured skin and very light brown or blue eyes.

The form OCA2 is the most frequent, particularly among black people from South Africa. It is characterized by activity of the tyrosinase enzyme that produces a photomelanin red-yellow pigment. This pigment gives the hair of the individuals affected a yellowish colour and makes the iris of their eyes light brown.

There is also a group of “unclassifiable albinism” for individuals with OCA who cannot be considered part of the three main groups, either because their phenotypic characteristics are blurred or because these are not present in the typical form.

In general the prevalence of the condition sees albinism as a relatively common phenomenon among the population of sub-Saharan Africa (from Nigeria to South Africa), especially in Zimbabwe and in other parts of southern Africa. An overall evaluation of the phenomenon of albinism shows an incidence ranging between 1 in 1500 and 1 in 15,000 individuals.

Ethnic origin undoubtedly plays a role in the prevalence of this genetic condition in Zimbabwe, which has the highest number of cases in southern Africa; most albinos (83%) belong to the Tonga tribe which, over the years, has had a limited number of migratory movements and resides exclusively in southern Africa.

Other factors also have to be considered, such as the maintenance of certain ritual matrimonial practices that accept marital unions between relatives. This favours the persistence of genes which carry albinism in the same geographical areas, facilitating transmission to future generations.

Materials and methods

In this article we examine two cases of two children that came to our attention in August 2009, a sister and brother affected by oculocutaneous albinism type 1 (negative tyrosinase).

The girl, aged 13, was born in French Guinea and at the age of 3 years moved with her family to Italy. The boy, aged 7, was born in Italy and is the penultimate of four brothers and sisters. The other two children show no signs of oculocutaneous albinism. Both parents are phenotypically normal and, according to the children’s father, there are no other cases of albinism in the family.

SUMMARY. Oculocutaneous albinism is an autosomal recessive disorder. It is associated with a disorder in the synthesis of melanin pigment, clearly manifested by the absence of colour in the skin, iris, and hair. In addition to its importance for a person’s physical appearance, melanin plays a protective role with regard to solar radiation; its absence exposes the skin to a greater risk of related photogenic injury. The two young patients reported, suffering from oculocutaneous albinism, developed first- and second-degree superficial burns after a few hours of exposure to the sun.

Keywords: oculocutaneous albinism, South Africa, burns, nystagmus, OCA

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In addition to pale skin and sandy hair, the two children show ocular alterations typical of albinism, i.e. spontaneous and continuous bilateral nystagmus and a reduced level of melanin in the iris (both children have blue eyes).

Melanocytes are normal in such individuals but the melanosomes contained within them do not reach a sufficient degree of maturity to allow them to deposit the correct amount of melanin.

The pale skin, without any protection owing to the lack of the melanin pigment, exposes patients to a higher risk of developing malignant skin lesions than normally pigmented individuals. The body parts at highest risk of developing skin damage are those that are most exposed: the face, ears, neck, and shoulders. The lesions that may occur include elastosis, solar keratosis, freckles, and superficial ulcers. In these cases exposure to sunlight is very harmful, as it can quickly cause major burns all over the body.11

The two children came to our attention for the treatment of extensive burns in most of their skin, due to excessive exposure to the sun one day at the seaside.

The children had been exposed to sunlight for about 5 hours, though not continuously, between mid-morning and mid-afternoon. They had had abundant sun cream spread all over their body before the exposure, but evidently this was not enough to prevent sunburn.

### Results

The two children were sent to us by the Emergency Department of Parma Hospital in Italy. The boy, aged 7, presented first-degree burns in the lower and upper limbs, abdomen, and chest. He also had superficial second-degree burns with small blisters, some of which had already broken and were partly drained by the nursing staff, on the shoulders, face (especially on the surface of the helix of both ears), and in the crease of the forearms. The burning sensation and discomfort that he experienced were clearly very marked.

We saw the children again after three days and re-
ferred them to our paediatric colleagues. They were told to perform their medication once a day only in areas of first-degree burns, to drink plenty of water, and to take paracetamol in the event of pain (Figs. 1a, b, c, d).

After three days we reassessed the burns, which had improved. There were no evident signs of infection, and the children did not feel as much pain as before.

The process of re-epithelialization had begun, but some discoloured areas in the patients’ skin where the burns were may remain. These areas were bright pink (Figs. 2a, b, c, d, e, f).

After the second medication the two children were discharged from the Department of Paediatrics. We recommended that they should continue daily washing of the burn areas with mild soap, using a moisturizing cream all over the body and carefully avoiding further exposure to the sun.

We agreed to see the two children again after two months to evaluate any possible developments (Figs. 3a, b, c, d, e, f).

Discussion

The two cases we describe were a valuable source of information for a study of the management of patients followed by our burns unit. Both cases were children of African origin.

Generally speaking, the skin of individuals belonging to this population has a high protective power against solar radiation. In our two cases, however, the young patients were born with a much lower level of protection than most individuals both in the same ethnic group and among Caucasians: the sun protection in these two children was almost non-existent.

Another feature that we had to consider is the tendency of blacks to develop keloidal scars much more frequently than other populations.

The children came through the re-epithelialization phase in optimal manner in a matter of months post-burn. There were no residual marks of any kind, and no hypo- or hyperchromic locations.
The skin lesions healed completely and despite the high probability of pathological healing, the skin of the two children showed no signs of any hypertrophic or keloidal activity. The children did not report any changes in sensitivity in the burn areas.

RÉSUMÉ. L’albinisme oculocutané est une maladie autosomique récessive associée à des troubles de la synthèse de mélanine qui se manifeste avec l’absence de couleur de la peau, de l’iris et des cheveux. A part l’aspect physique, la mélanine joue un rôle protecteur contre le rayonnement solaire: son absence expose la peau à un risque majeur de lésions cutanées photogéniques corrélées. Les deux jeunes patients étudiés, atteints d’albinisme oculocutané, avaient subi des brûlures superficielles de premier et deuxième degré après quelques heures d’exposition au soleil.

Mots-clés: albinisme oculocutané, Afrique du Sud, brûlures, nystagmus, AOC


