Albinism is the name given to a group of inherited disorders characterized by a defect in melanin production clinically leading to little or no pigmentation in the skin, hair and eyes. There are a number of genetic abnormalities that can produce this problem, the majority also significantly affect the eye and are also known as oculocutaneous albinism. This article describes the clinical features, incidence and cause of this condition and also outlines the impact that this condition has on persons affected with albinism (PAA) with a worldwide perspective and a particular focus on approaches and initiatives in dealing with this in resource poor settings of the developing world.

**Key words**
Oculocutaneous albinism  
Chediak-Higashi syndrome  
Hermansky-Pudlak syndrome  
Griscelli syndrome

Oculocutaneous albinism is an inherited disease that is found worldwide. However, it is in the tropical parts of the world where the clinical and life-altering impact is felt most predominantly by persons affected with albinism (PAA) due to sun exposure and skin cancer, social exclusion and persecution secondary to lack of understanding of the cause and nature of the condition.

How common is albinism?
Although its overall global prevalence is reported to be 1/20,000 of the population (Witkop, 1989), it is more common in parts of Africa (Hong, 2006) with a prevalence as high as 1/1000 in the Tonga tribe in Zimbabwe (Lund, 1997).

The cause of albinism
Albinism represents a group of pigmentary disorders where production of melanin is impaired for some reason. There are many different mechanisms for this but one example, oculocutaneous albinism type 1 occurs due to an absent gene (1A) or due to a defective gene (1B) producing tyrosinase, an enzyme involved in the melanin pathway. The majority affect the skin and the eye and are known as oculocutaneous albinism with just one variant of albinism confined to the eye (ocular albinism). People who have ocular albinism have generally normal skin and hair colour, and may even have a normal eye appearance.

The oculocutaneous group can be further divided according to whether they have the enzyme tyrosinase, which is involved in the production of melanin. For example, OCA-1a is tyrosinase negative and OCA-2 is tyrosinase positive. However, things are not quite as simple as this now with advances in molecular genetic techniques and there are many different types of albinism, classified according to the identification of the genes responsible. Some of the different types of albinism include:

- Oculocutaneous albinism:  
  - type 1  
  - type 2  
  - type 3  
  - type 4

- Ocular albinism:  
  - x-linked ocular albinism is also called Nettleship-Falls ocular albinism  
  - ocular albinism with sensorineural deafness  
  - Chediak-Higashi syndrome  
  - Hermansky-Pudlak syndrome  
  - Griscelli syndrome.

Inheritance is mainly autosomal recessive and there is one form, associated with deafness, that is x-linked.

Recognising albinism
In all racial groups there is a marked reduction in the intensity of the pigmentation of the skin, hair and eyes. In OCA-1 (tyrosinase negative), the most severe form of the disease, the skin is pink, hair white and the eye colour pink, showing a marked red reflex when the eyes are examined. In OCA-2 (tyrosinase positive), a small amount of pigment develops with the hair becoming yellow/brown and the skin developing large freckles or solar lentigines, especially on sun-exposed sites.

The eye features are striking. All groups typically experience photophobia, discomfort in bright light. Nystagmus (involuntary rapid movement of the eyes resulting in them oscillating from side to side or round and round) either horizontal or rotatory, is possibly the most striking feature. Refractory problems also occur and many children...
The social impact of albinism is significant, with social exclusion being not uncommon. In developing countries there are many superstitions surrounding the cause of the disease. These often convey negative connotations, such as a curse having been put on the family, misbehaviour of the mother, or derogatory name-calling such as zeru zeru (ghost), mzungu (white person), and ‘pig’ (McBride and Leppard, 2002).

While over 50% of people with albinism will have an affected relative, the majority of native Africans do not understand that it is an inherited disease, and attributing the disorder to a spiritual curse is the most common misconception. The birth of a child with albinism can have a severe psychological impact on the parents fuelled by some of the myths surrounding albinos and the unusual, striking and contrasting white skin colour in black races. Work from South Africa (Kromberg, 1984) showed that the normal bonding process between mother and child is delayed and may take up to nine months postnatally to develop fully. Although work shows that people with albinism are socially well integrated, they tend to remain unmarried (Kromberg, 1984). Even in 2008, reports from Tanzania in the international press describe how albinos are being killed for their body parts (Allen, 2008).

Measures to address these issues are being implemented using educational and other resources, such as those included in the Albino Outreach programme in Moshi, Tanzania. This incorporates an albino outreach clinic designed to educate and prevent the development of solar damage through the distribution of sunscreens and protective clothing, education of the teachers, provision of magnification boards for school children, and management of solar-related pathology through regular dermatological check-ups (Kromberg, 1984).

Treatment for albinism
While there is no treatment to correct the underlying lack of pigment, much can be done to support the individual. A key start is to make the diagnosis early and instigate the necessary support network and preventative care as soon as possible. In temperate climes, correcting refractory eye problems with appropriate lenses ensures that the child progresses at school, achieving the key reading skills for life. Sun protection is an issue and can be managed relatively easily with photo protection and limiting exposure.

Genetic counselling should be undertaken to enable education regarding inheritance of the condition and the risks to subsequent progeny. Although work in South Africa suggests that families were unlikely to limit their family size on understanding the 1:4 risk of having a further PAA, although, of course, they
would prefer the next baby to be normal. While prenatal diagnosis is theoretically available, it is not practically so to the majority of mothers pregnant with a potential PAA (Eady et al, 1983).

In the tropics, management of this disorder is complex and multifactorial. It is a public health concern in sunny climes, especially developing countries where PAA are at increased risk of the development of and premature death from skin cancer and sun damage. This is for several reasons:

- If refractive errors are not picked up and resolved they lead to difficulties at school, resulting in the child leaving without the ability to read and write. Thus, many PAA are forced to take on unskilled work and, in tropical Africa, this generally involves working in the fields, thus spending the heat of the day under bright sunshine potentiating early photo damage.
- Chronic sunburn leads to the early onset of chronic sun damage of the skin with solar elastosis. This may be widespread in children as young as eight.
- Chronic sun damage results in the early onset of skin cancer; especially squamous cell carcinoma (SCC).
- In developing countries, where intertribal marriages lead to an increased expression of genetic disorders, albinism is more common (Okoro, 1975).
- Health infrastructures in developing countries may not be as well formed so facilities for health education and prevention, as well as services and expertise available for managing skin cancer, is limited.

Public health initiatives
An example of an excellent public health approach to managing PAA comes from the foothills of Kilimanjaro in Tanzania. Here, the albino programme was set up more than 10 years ago. Dr Alfred Naburi and Mr Peter Kundy continue to run the service out of the Regional Dermatology Training Centre, Kilimanjaro Christian Medical College (KCMC), Moshi, Tanzania (Figure 1). This programme consists of:

- Establishment of a dedicated team lead by a co-ordinator (Peter Kundy) and clinical leader (Dr Alfred Naburi). This team administer and run monthly clinics, raise awareness and dispel myths in communities, especially in schools and among churches and mosques.
- Supporting PAA in schools, including the provision of reading aids and educating the teachers of the special needs of these children.
- Advocacy both locally, nationally and internationally (Hibbert, 2006), thereby raising awareness of the true nature of the disorder and dispelling the commonly held harmful myths.

This programme now runs outreach clinics in at least six surrounding localities enabling PAA in the area to be seen twice per year. At these clinics sun avoidance measures are reinforced, donated sun screen is distributed and support for the provision of suitable clothing, i.e. hats, long sleeves and long trousers is given. The skin is checked for any signs of skin cancers or pre-malignant conditions and simple lesions may be treated on site with liquid nitrogen. For more complex lesions, the patients are invited back to the hospital for surgery.

Conclusion
Albinism is a worldwide genetic disorder resulting in absence of the capacity to produce normal cutaneous and ocular pigmentation. While in temperate countries the health consequences of this are, in the main, secondary to the eye problems, and correctable as well as cosmetic, in sunny tropical climes the outcomes can be dire without adequate early sun protection measures and timely intervention for the early management of skin cancers.

Much can be done to support PAA. In Tanzania we see an excellent functioning model of outreach care which is run at relatively low cost and is locally sensitive and relevant.

Key points
- Albinism is an inherited disorder.
- It is caused by a number of different gene defects.
- PAA have little or no melanin in the skin.
- PAA are vulnerable to sun damage, sun burn and the premature development of skin cancer.
- Community-based clinics such as the Albino Outreach Programme in Moshi, Tanzania provide local, relevant and effective support for PAA in outreach settings.

Hibbert K (2006) In the shadow of the sun. Sunday Times, July 9th

References

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