



Understanding Albinism

What is Albinism?

People with Albinism are born with little or no pigment in their eyes, skin and hair, although in some cases the reduced pigmentation only affects the eyes. Albinism occurs because people have inherited genes that do not produce the usual amounts of a pigment called Melanin.

In the United Kingdom, one in 17 000 people are born with some type of Albinism. Albinism occurs in all races and most children with Albinism are born to parents who have the hair and eye colour that would be for their ethnic background.

There are two main types of Albinism:

Oculocutaneous Albinism involves the eyes, hair and skin. Within this type of Albinism, researchers have to date identified ten different variations.

Ocular Albinism involves primarily the eyes, although some people with Ocular Albinism may have slightly lighter skin and hair compared with other family members.

Why do people have albinism?

Albinism is referred to as an autosomal recessive condition. It is passed from parents to their children through genes. In almost all types of Albinism (not Ocular Albinism), both parents must carry an Albinism gene to have a child with Albinism. Many parents carrying the Albinism gene will have normal pigmentation. When both parents carry the gene and neither parent has Albinism, there is a one in four chance at each pregnancy that the child will be born with Albinism.

How does Albinism affect people?

Those born with Oculocutaneous Albinism, the most common type, are very sensitive to light, have very pale blue or blue/grey eyes and little or no pigment in their skin and hair. Many will wear tinted spectacles or contact lenses to reduce discomfort from glare.

People with type 1 Oculocutaneous Albinism, also referred to as Tyrosinase Negative Albinism, will have no pigmentation at all in their hair and skin. The hair will generally appear white or platinum coloured and the skin will be very pale. The eyes will appear pale blue or blue/grey and most will have a significant visual impairment (a partial sight).

People with type 2 Oculocutaneous Albinism, also referred to as Tyrosinase Positive Albinism, will have a lack of pigmentation in their skin and hair. However, this will not normally be as marked as with type 1, as they will have some visible pigmentation. Generally people with this type of Albinism will have pale blonde hair and

blue or sometimes light brown eyes. Due to the increased level of pigmentation, the vision of people with this type of Albinism will not normally be so severely affected.

People with Ocular Albinism will generally not display a significantly reduced level of pigmentation although the eye problems associated with Albinism will be present.

In addition to the main types of Albinism mentioned above, there is also a rare type called Hermansky Pudlak Syndrome (HPS). This type of Albinism can involve a variety of complications associated with bleeding and respiratory disorders. It is essential that a child's type of Albinism is identified early in life. HPS is extremely rare, but if a child appears to bruise very easily or has any bleeding difficulties HPS should be suspected and specialist advice should be sought.

Effects on the eyes?

The most important thing to remember about Albinism is that the worst the vision can be is on the first day of life. That is to say that Albinism is a static condition. Albinism generally affects the eyes in a number of ways:

Nystagmus

This is an involuntary movement of the eyes, usually from side to side but sometimes also up and down. The brain does however adapt to the movement and so whilst the eyes might move, the image a person with Albinism is seeing does not.

Strabismus

This means that there is a muscle imbalance within the eyes causing the person to have a squint and also to use each eye separately rather than together. Thus people with Albinism do not have binocular vision (the ability to use both eyes at the same time). Again however most people adapt and so there is not a sense of double vision.

Photophobia

Most people with Albinism will be very uncomfortable in bright light due to an absence of pigment in the eyes. Too much light can enter the eye creating glare and discomfort. Most people overcome this difficulty through the use of tinted spectacles or contact lenses. Wearing a peaked cap can also help reduce glare.

Other Eye Information

People with Albinism may also be far sighted or near sighted and can also have astigmatism. This is where the surface of the eye is not spherical and therefore there is a distortion in the image seen by the person. These aspects of the condition can often be helped by wearing prescription spectacles or contact lenses.

These eye problems result from abnormal development of the eye because of lack of pigment. The retina – the surface inside the eye that receives light – does not develop properly before birth and in infancy, so that vision is not sharp even with corrective lenses. The nerve signals from the retina to the brain do not follow the usual nerve routes. The iris – the coloured area in

the centre of the eye – does not have enough pigment to screen out stray light coming into the eye. Light normally enters the eye through the pupil – the dark opening in the centre of the iris – but in Albinism light can pass through the iris as well.

Many people with Albinism can and do register as partially sighted or blind. These terms are medical and they refer to how much a person is able to see on an eye test chart. It is important to remember that the level of vision which the person with Albinism has is entirely normal to them.

How can people with Albinism maximise their sight?

Low Vision Aids

Many people with Albinism are able to benefit from a wide variety of low vision aids LVAs such as hand held magnifiers for reading small print; monocular or telescope for viewing distant objects; and a range of electronic aids such as video magnifiers (also know as closed circuit televisions - CCTVs).

Most people with Albinism will be able to access computers with the help of a large monitor and/or software which enlarges the display or converts it into speech. Many people with Albinism are encouraged to try out different types of tinted lenses to see which offers the most comfortable level of protection from glare.

It is essential that children with Albinism are seen by an Ophthalmologist regularly so that they have an appropriate spectacle or contact lens prescription.

Skin Issues

People with Albinism need to take care of their skin to avoid the risk of burning in any weather conditions. Due to a lack of pigment, many people with Albinism will be aware that their skin burns easily and so it is recommended that a high factor UV block sunscreen be worn on all exposed areas of skin.

People with Albinism are at an increased level of risk of developing skin cancer so it is essential that a good skin care regime is maintained from an early age. A hat with a peak is a very useful aid for many people because it can offer sun protection and a good means of shading the eyes from bright light. Children should be encouraged to wear a variety of hats when outdoors. These should be fashionable and attractive to the child so that he/she is happy to wear their hat.

Social Issues

Some people with Albinism may experience torment at school through name calling and bullying. It is essential that this is checked and reported at the earliest opportunity. Children should be encouraged to be open about their Albinism and it would be very beneficial for them to have contact with others with the condition as a means of encouraging good self esteem.

Apart from the vision issues outlined above and the need to ensure protection from the sun, most people with Albinism live normal lives and participate in the same range of activities as everyone else. Certain sports involving a small fast moving ball may prove difficult for people with Albinism. However it is

essential that children are encouraged to try as many activities as possible and that they are given the opportunity to find out what sports they enjoy.

Many hobbies and interests are possible with the use of an appropriate simple low vision aid such as a monocular for watching football or rugby. It is also important to think about contrast. By using a brightly coloured ball for example it is often possible for people with Albinism to take part in a range of ball games.

Many people including those with Albinism can benefit from making use of colour in the choice of their clothing. Certain colours can be used to enhance skin tone and hair colour thus enabling an individual to make the most of their appearance and feel good about themselves. Young people should be encouraged to experiment with colours and achieve a feel good look.

Albinism Fellowship

Albinism Fellowship is a support organisation for people with Albinism in the UK and Ireland. Founded in 1979 the fellowship is a purely voluntary organisation which offers advice and support to people with Albinism, their parents and families, and others with an interest in the condition.

Albinism Fellowship aims to raise positive awareness of the condition by providing opportunities for contact with other individuals and families. A quarterly newsletter, telephone helpline, website and bi-annual conference are among the current services on offer to members.

The fellowship is a sociable organisation and many members find that the opportunity to develop and maintain contacts with others with Albinism is invaluable.

Further information on Albinism and Albinism Fellowship can be obtained from:

**Albinism Fellowship
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E-Mail: support@albinism.org.uk
Web: www.albinism.org.uk**

The US organisation, NOAH also has a website with useful information at www.albinism.org