Albinism

Radhika Holmström looks at a complex and often misunderstood genetic condition, and asks whether society's attitudes to albinism are worse than the condition itself.

"Albinism is much more common than people think, but it's far under the radar in terms of funding, research and public awareness", says Dr Glen Jeffery, who is the professor of neuroscience at University College London's Institute of Ophthalmology, and one of the few specialists in the condition. In fact, many people don't realise that albinism results in very poor vision – and/or that this is virtually the only disabling condition it causes.

People with albinism are subjected to a rather different kind of treatment than many other disabilities: and to the casual observer, it's the other – non-disabling – aspects that are considered noteworthy, rather than the actual disability which results. As Robin Spinks of the Albinism Foundation puts it, "It combines vision problems with visual distinctiveness – and it's usually misunderstood."

The myths

In many cultures, that misunderstanding is expressed in genuinely terrifying ways. In Tanzania alone, people with albinism are regularly murdered for the use of their body-parts in traditional medicine – and until recently such murderers had never been convicted. Journalist Rob Curran described a couple of years ago how "when I went to live in South Africa for two years, I realised that the treatment I'd received... was minor in comparison to how black people with albinism are treated" and how "in Zimbabwe, Tanzania, Lesotho and other African countries, in many rural areas there are beliefs that people with albinism are cursed and are mentally sub-normal. In Zimbabwe there is an almost complete absence of people with albinism in the catering industry as there is a widespread misconception that albinism is infectious. A man called Milton said that in his home, on the Eastern Cape, he was told by village elders that he would never die, but would simply disappear. Thrown out of his village at the age of 19, with no qualifications, he told me how there was a belief among young men with HIV that raping an albino woman would 'cure' them."

In the West, the prejudices are less lethal; but most people with albinism are subjected to a quite extraordinary level of misconception and abuse on a daily basis. There is also a level of unhelpful fascination in the physical

appearance of people with albinism, with villainous depictions in popular culture being common – from Silas, the evil monk in 'The Da Vinci Code', to the 'Twins' in 'The Matrix reloaded'. The Albinism Fellowship is probably unique among disability organisations in needing to state explicitly on its website "Please note that we are NOT an agency for hiring, casting or modelling".

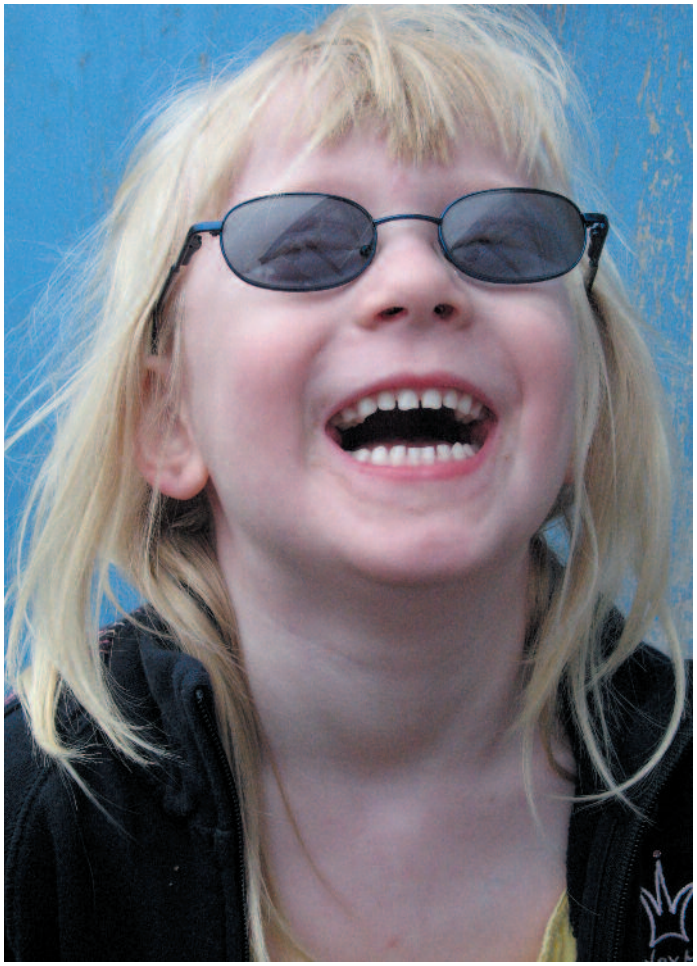
The reality

Nor does everyone with albinism have the stereotypical white skin and white hair with mauve eyes. The condition has a number of different variants (depending on which gene mutation has caused it) – and 'hypopigmentation' (where the skin becomes lighter because of problems with melanin production) in even more. In fact the US equivalent of the Albinism Fellowship is the National Organization for Albinism and Hypopigmentation. But broadly there are two categories, ocular albinism (OA) and oculo-cutaneous albinism (OCA), with variants within these.

People with OA do not necessarily look much paler than those around them, especially if they are in Northern Europe, but their eyes are affected. OCA is much more noticeable, but even some people in this group have red or brown hair, and some pigment in their skin so that they do tan. The estimate most commonly used is one in 17,000 children in the UK. In other cultures it may be more or less common, depending partly on whether close relatives produce children together; but Jeffery points out that it may well be substantially higher in the UK too. "In reality, if we include people who have hypopgimentation problems, it's probably three to four times higher than the estimate."

The different forms of albinism are inherited, and diagnosis can be complex. The gene for OA is linked to the X chromosome, which means that women (who have two X chromosomes) may only be slightly affected if they have it on one X chromosome but will be 'carriers'. Their sons – who have only one X chromosome – thus have a 50 per cent chance of having full OA. OCA, on the other hand, is inherited in an 'autosomal recessive' way, which means that both parents need to carry a particular gene mutation for the condition to occur in the child and there is a 25 per cent chance that any pregnancy will result in a child with OCA.

"The clinical diagnosis can be quite difficult to do", adds geneticist Dr Patricia Lund. "For instance there are at least four genes involved in OCA. OCA1 'codes' for the key enzyme tyrosinase – if that enzyme is knocked out you get very low levels of pigment – but you can get all sorts of mutations throughout the genes. There's no predominant one. ➜



"OCA2 is linked to a different gene, and there's not a total loss of pigment. And the children of one parent with OCA1 and one with OCA2 could be unaffected, because of the way the condition is inherited."

There are also a few rare variants of OCA, which are syndromes linked to other health and/or disabling conditions. Otherwise, the main effects are a much greater vulnerability to skin cancer, and on the vision.

Effects on the vision

Another common misconception about albinism is that the vision is damaged solely because there is no pigment in the eye to protect it from light. Certainly, people with albinism are sensitive to light and glare; but the main damage is done much earlier, in the womb.

"There are a lot of different genetic causes for albinism, but the result is that pigment – the melanin itself – is missing", Jeffery explains. "When you're developing and your cells are dividing to make a retina, they go through the 'cell cycle' in which they develop. In albinism, this is disrupted because one of the elements that you use to make pigment, dopa, regulates the cell cycle. No pigment means no dopa, and that means disruption to normal patterns of cell production. Dopa is the brake on how often they divide; in a normally pigmented embryo, cell production will stop at the right point, whereas in one with albinism production will carry on longer than it should. What then happens is that the retina gets too thick and there is a great wave of cell deaths. Lots of cells in the albino retina consequently die."

As a result, the fovea – the part of the retina with the highest number of photoreceptors – does not develop properly, meaning that vision is much less acute. In addition, says Jeffery, there are neurological consequences. "Your eyes connect to the brain differently. Normally, half of one's right eye projects to the same side, the other half the other way; in other words, each eye projects symmetrically to both sides of the brain. In albinism, the vast majority of the fibres cross to the other side of the brain. You have no binocular vision."

People with albinism also experience nystagmus (rapid movement of the eyes from side to side). Importantly, however, the brain adapts to this – and often to the lack of binocular vision – very well. And in fact, as Spinks explains, many people who have very poor vision or are indeed registered blind don't consider themselves "in the constituency that an organisation like RNIB works with".

Albinism in brief

- People with albinism are physically very distinctive because of their appearance. However, the only genuine disability caused by the condition is poor vision.
- The prejudices about albinism can be terrifying. In some countries, people with albinism have been murdered; others are thrown out of their homes and believed to be cursed.
- There's a fascination in popular culture with the physical appearance of people with albinism – often depicting them as figures of evil.
- In reality, albinism has many different variants, depending on the gene mutation responsible. There are two main categories: ocular albinism (OA) and oculo-cutaneous albinism (OCA).
- People with OA do not necessarily look much paler than those around them, especially if they are in Northern Europe; but their eyes are affected. OCA is much more noticeable, but even some people in this group have red or brown hair, and some pigment in their skin so that they do tan.
- The estimate most commonly used is one in 17,000 children in the UK, although some specialists argue that it should be much higher. In other cultures it may be more or less common, depending partly on whether close relatives produce children together.
- The gene for OA is linked to the X chromosome. OCA is 'autosomal recessive', which means that both parents need to carry the gene mutation for a child to show symptoms and there is a 25 per cent chance that that any pregnancy will result in a child with OCA.
- Albinism principally affects the vision because the retina cannot develop properly in utero without pigment. Without pigment to trigger the right cell development, the retina gets too thick and then a lot of cells die off.
- The part of the retina responsible for the most acute vision does not develop. The eyes also connect to the brain differently, which means no binocular vision. Nystagmus (constantly 'wobbling eyes') also results. However, the vision does not deteriorate and many people do not consider it a major problem even if they are registered blind.
- There is no way of repairing the damage to the eye and brain. Low vision aids can help the vision problem; but the main problem is with other people's perceptions.

He elaborates: "Vision is normal to them – there is no sense of deficit or loss, because the worst the vision can be is on the first day of life. It's a stable condition, which won't deteriorate until the stage in one's life that everyone's vision deteriorates."

Treatments

The only 'treatment' for albinism is the conventional range of low vision aids on offer. More generally, Jeffrey stresses, there is nothing that can be done: the groundbreaking work on retinal repair and/or gene therapy is not applicable here. "The damage is too fundamental. Repairing it would require major architectural changes to the eye and brain.

The important thing is to tackle the myths around the condition. Your average ophthalmic consultant tends not to know much about it; but it's a perfectly stable condition which won't get any worse and won't get any better – except that in practice, in fact, it will get better because from babyhood on, people learn strategies for coping with it."

Everyone involved with albinism agrees that it's the myths that need tackling, not the condition itself. In many ways, in fact, it's a perfect demonstration of the social model of disability: that it is society, not medical conditions, that disable people. The biggest barriers anyone with albinism faces are other people's perceptions, not the limits on physical or sensory ability associated with albinism itself.

The images in this article are taken from 'Real lives: Personal and photographic perspectives on albinism' by Archie W N Roy and Robin Spinks, with photographs by Rick Guidotti. Published by the Albinism Fellowship, price £12 including post and packing.

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