



Vision

Keratoconus and Down's syndrome

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Keratoconus is a condition in which the cornea, the transparent 'window' at the front of the eye, stretches into a cone shape, causing distorted vision. It usually begins in the teenage years through to mid-twenties. This condition occurs in the general population (about 1 in a 1,000) but is much more common in young people with Down's syndrome, and may affect as many as 1 in 10. At the moment, we can't predict which young people will be affected and young people who have never had any eye problems are as likely to get keratoconus as are young people who have always worn glasses.

Until recently, there was no treatment for early stages of keratoconus and nothing to be done about it until vision was affected, when contact lenses can be a real help. In cases of severe progression, scarring of the cornea meant that a corneal transplant was the only means of providing reasonable vision. Now, there is a new treatment becoming available, called collagen cross-linking therapy that can halt the progress of the abnormal growth and prevent sight deteriorating, if caught in the early stages. It works by soaking vitamin B into the front of the cornea and exposing it to UV light. This stiffens the cornea to prevent the shape from deteriorating. For many people (including people with Down's syndrome), the procedure is carried out under local anaesthetic. Since the patient needs to keep their eyes very still for up to 30 minutes, a general anaesthetic is more suitable for some patients. Cross-linking is not yet available in all UK NHS eye departments, but is available at many regional centres throughout the UK and patients can be referred to one of those centres for treatment.

Before cross-linking, there was no urgency in diagnosing keratoconus in its early stages, because patients were simply treated with glasses or contact lenses and neither prevented the condition progressing. However, cross-linking therapy is only viable in relatively early cases, so it is really important that the condition is picked up as early as possible. However, there are challenges for young people with Down's syndrome:

- People with Down's syndrome are less likely to report changes in their vision, so changes may go un-noticed by family and friends
- Cross-linking can be used only when the cornea is at least 375 microns thick (that's 0.375 millimetres). In the general population, the healthy cornea is about 550 microns thick, but in people with Down's syndrome, the healthy cornea is much thinner, on average about 475 microns. This means that there is a much shorter time window for keratoconus to be picked up and therapy initiated in young people with Down's syndrome, before it's too late.
- Families of young people with Down's syndrome, especially those who have never needed to wear spectacles, are unaware of the possibility of keratoconus and the importance of regular eye examinations.

In Cardiff, we have been conducting a research study to determine the best way of picking up keratoconus in the earliest stages. The defining features of the condition, i.e. the abnormal shape of the cornea is diagnosed by an instrument specially designed for the purpose; a corneal topographer. This is not available in all optometry practices and even when it is available, young people with Down's syndrome find it very difficult to co-operate with its use. We wanted to find reliable tests that are quick, simple and acceptable to young people with Down's syndrome. We measured a number of visual functions and characteristics of the eyes in a group of 45 young people with Down's syndrome, aged 13 to 26 years, 11 of whom had keratoconus. Our results were:

- Eye-rubbing (long held to be a 'cause' of keratoconus in Down's syndrome) does NOT distinguish those individuals at risk of keratoconus and does not seem to be a cause of keratoconus at this stage
- Measuring astigmatism (associated with keratoconus in the general population) does NOT distinguish those individuals at risk of keratoconus
- Measuring visual acuity (the standard letter or picture chart) does NOT distinguish between healthy eyes and early keratoconus
- Examining the health of the exterior eye under magnification does NOT distinguish between healthy eyes and early keratoconus
- The quality of the retinoscopy reflex DOES distinguish between healthy eyes and early keratoconus

A retinoscope is a specialised hand-held 'torch' used to shine light into a patient's eyes and the retinoscopy reflex is the light reflected back from the eye, observed by the optometrist. We are all familiar with cats' eyes shining in the dark; human eyes reflect light too, but in smaller amounts, usually only noticeable as 'red-eye' in flash photographs. The retinoscopy reflex can be used to judge the eyes' focusing and determine a spectacle prescription. In keratoconus, the light forming the reflex is less even and more 'patchy' in brightness, even in very early stages of the condition. In our study ALL eyes with keratoconus had an abnormal retinoscopy reflex and ALL healthy eyes had a smooth reflex. So this technique, which all optometrists are trained to use, is the obvious key to detecting keratoconus in young people with Down's syndrome.

However, there is a catch. Just as in many spheres of life, practitioners are being seduced by technology and in many optometric practices, the retinoscope is being replaced by 'auto-

refractors', machines that automatically measure a patient's spectacle prescription at the touch of a button and without the practitioner viewing the patient's eyes directly. Auto-refractors are usually used by non-professional staff and the results handed on to the optometrist. Early keratoconus can pass unnoticed, or difficulty in obtaining a result with an auto-refractor put down to the patient's learning disability. Optometrists that rely on auto-refractors are losing skills that are vital to good health care for people with Down's syndrome.

The message for parents then, is this: ensure that your child / young person has an annual eye examination, with a practitioner that not only understands the communication needs of people with Down's syndrome, but that also is highly skilled in retinoscopy and knows the risk of keratoconus in this group. How are you to go about ensuring that? Ask the following questions:

- Does the optometrist regularly see children and adults with Down's syndrome?
- Does the optometrist use retinoscopy with ALL patients, not just those with learning disabilities?
- Is the optometrist familiar with the visual problems of people with Down's syndrome?
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If the answer is yes to all three, then make the appointment, otherwise go elsewhere. During the appointment ask direct questions about assessing your child / young person for keratoconus. If you do not have confidence in the answers, don't be afraid to walk away.

Does your child or a young person you support have a diagnosis of keratoconus?

If they are referred to the hospital, ensure they get the opportunity to have regular topography scans to understand the shape of the cornea and to learn if it is stable in shape, or if it is changing over time. Results can be variable, so it's important that your young person is given lots of practice with the machine and are not rushed. Several scans should be taken so that the average can be used.

Find out early if they are suitable for having cross-linking treatment.

Ensure that they can try spectacles or contact lenses to improve their vision. Having Down's syndrome can make contact lens wear more challenging, but it depends upon the individual themselves.

Finally, if the keratoconus is severe, ensure that you discuss with the doctor surgical options such as intra-corneal ring segments to reshape the cornea or a corneal transplant to replace scarred tissue. Surgery such as transplant is not an easy-fix and does not guarantee success, requiring intense care using drops for a few weeks afterwards, and many visits to the eye department. However, it is important that your young person's perceived options are not limited due to the perception of their disability and that their visual needs are carefully considered.

www.keratoconus-group.org.uk and lightforsight.org are both a great source of further information and support for those with a diagnosis of keratoconus.