

# Australian Study of Keratoconus (ASK)

March 2014

**Keratoconus** is a disease in which the cornea (the clear front window of the eye) progressively thins and begins to protrude, resulting in irregular astigmatism and decreased vision. Despite much research, keratoconus remains a poorly understood disease. At the Centre for Eye Research Australia, we are undertaking an extensive “Australian Study of Keratoconus” to better understand keratoconus and the role that genetic and environmental risk factors play in the disease. To date, we have recruited 300 keratoconus patients and we would like to thank all the participants who have already donated their time and been involved in this study. We have undertaken an initial analysis of these data and we present some of the results below:

## Association with allergy

Our results showed that *asthma* as well as *eczema* appeared as significant risk factors for keratoconus. Going forward, investigation of the association between these 2 conditions and keratoconus needs to be clarified as to whether they share common elements.



## Changes on imaging the cornea

Imaging of the cornea has demonstrated that the thickness of the cornea at different locations is an important parameter for detecting subclinical keratoconus (disease that shows no clinical signs and symptoms) and that the cornea thins with increasing disease severity. We would like to perform further clinical examinations of family members of individuals with keratoconus to assess if a thinner cornea is inherited which may thus present as an underlying risk factor. As topographic imaging techniques have improved, the number of close relatives of affected individuals able to be identified with some change in their cornea has increased from 6–8% of subjects to 50%. Thus better imaging is leading to increased disease detection and the possibility of early treatment interventions.



Figure of the Pentacam imaging system used for analyzing the front of the eye.

### Changes to the back of the eye

We have identified that certain changes at the back of the eye ('retina') are also present in keratoconus eyes compared to non keratoconus eyes. However we do not know if these changes occur prior to, or as a result of keratoconus. Further study is required to establish the clinical implications of these findings. Such changes appear to occur in early disease and thus it would be of interest to assess members of keratoconus families who are currently undiagnosed with disease. Imaging of the retina, in addition to the corneal imaging, may provide a possible diagnostic test in individuals with keratoconus before it is clinically apparent thus allowing treatment options to be considered at the earliest possible stage.

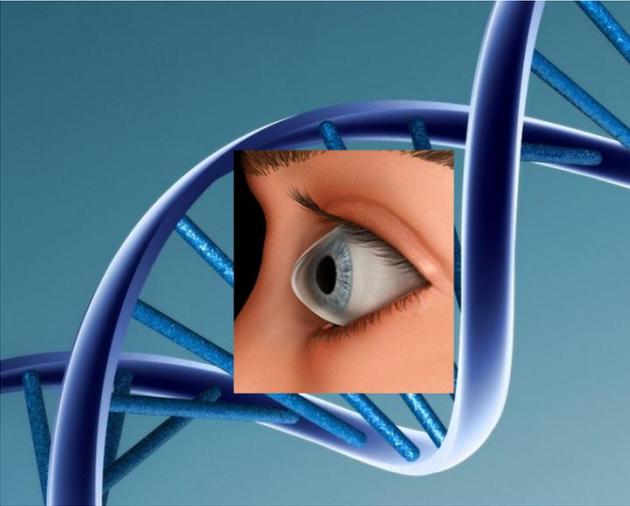
### Quality of life

Having noticed considerable changes in various measurements of keratoconus eyes, we assessed the impact of keratoconus on vision-related quality of life in adults. The study demonstrated that worse vision in the better eye (but not the worst eye) was independently associated with a reduction in vision and quality of life. This suggested that the better eye is a more important factor on the impact of keratoconus from the patient's perspective. Further investigation of the changes in quality of life over time, and before and after cross linking or corneal transplantation, will provide ophthalmologists with a better understanding of the impact of management plans from a patient's perspective. Also there is a lack of information on the impact of keratoconus on quality of life in children. Therefore we would like to assess vision and quality of life in children as this would provide useful information to assist in planning a child's support needs.



### Genetics

Genetic studies have assessed a number of possible genes and several have been implicated. One of these genes is called the hepatocyte growth factor gene and is known to be an important signaling gene in the body. Several corneal thickness genes have also been implicated and would therefore link in with the clinical observation of a thin cornea being a risk factor for keratoconus. These findings confirm that a portion of keratoconus has a genetic basis. Further studies on families of keratoconus patients to assess how these genetic variants occur in families will allow a better understanding of their role in the disease. A fuller knowledge of the role of genetics will assist in the early and accurate diagnosis of keratoconus and may pave a way for the development of new therapeutics to perhaps slow or ameliorate disease.



The availability of these preliminary data, allows us to target specific areas for further research and we now plan to recruit more keratoconus patients, children with keratoconus and families of individuals with keratoconus to better understand the causes of this disease. We also wish to evaluate the economic cost of the disease burden to individuals, as well as the costs of treatment relating to corneal transplantation and cross-linking.

We would like to take this opportunity to invite volunteers with keratoconus and their family members to participate in our studies. If you agree to take part in this study you will be invited to undergo an eye examination and donate a small blood sample at the Royal Victorian Eye and Ear Hospital, East Melbourne. Your help will allow us to advance our understanding of keratoconus as well as helping others.

### Publications

1) Evaluating the association between keratoconus and the corneal thickness genes in an independent Australian population. Sahebjada S, Schache M, Richardson AJ, Snibson G, MacGregor S, Daniell M, et al. Invest Ophthalmol Vis Sci. 2013 Dec;54(13):8224-8.

2) Impact of keratoconus in the better eye and the worse eye on vision-related quality of

life. Sahebjada S, Fenwick EK, Xie J, Snibson GR, Daniell MD, Baird PN. Invest Ophthalmol Vis Sci. 2014 Jan;55(1):412-6.

3) Association of the hepatocyte growth factor gene with keratoconus in an Australian population. Sahebjada S, Schache M, Richardson AJ, Snibson G, Daniell M, Baird PN. PLoS One. 2014;Jan.

### Media coverage

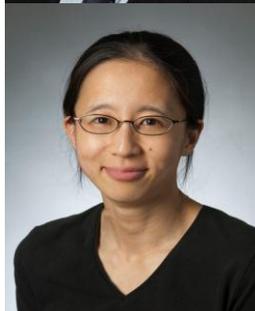
- Australian Optometry Journal - "Updates on Keratoconus", 2012.
- Vision Australia - "Updates on Keratoconus to general public", 2012.
- Centre for Eye Research Australia - "Sights set on a cure", 2011.
- Mivision Australia - "Myopia & Keratoconus", 2011.
- Dr Srujana Sahebjada profiled on cover page for Vision news Australia, 2011.

## Researchers involved in the project

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**Mark Daniell** is the Head of the Corneal Unit at the Royal Victorian Eye and Ear Hospital and Associate Professor at the Centre for Eye Research Australia and University of Melbourne.



**Dr Elsie Chan** is a Honorary Fellow at CERA and Consultant Ophthalmologist at the Royal Victorian Eye and Ear Hospital.



**Associate Professor Paul Baird**, is a molecular geneticist and heads the Ocular Genetics Unit at the Centre for Eye Research Australia.



**Dr Srujana Sahebjada**, is a Research Optometrist & Postdoctoral Fellow at the Centre for Eye Research Australia.

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