

# Diagnosing Keratoconus and Patients at Risk

Topography and adjunctive testing combined with the clinician's judgment are the keys.

BY YARON S. RABINOWITZ, MD

**G**ood clinical judgment combined with topography and adjunctive testing are the keys to diagnosing keratoconic subtypes. Understanding the spectrum of normal corneal topography is crucial to suspecting pathology.

## DEFINITIONS

I have developed a classification scheme based on anterior videokeratography and clinical signs to detect keratoconic subtypes that I hope will be useful to practicing clinicians. Keratoconus is a clinical disease that is detectable at the slit lamp through obvious clinical signs such as stromal thinning and is associated with a typical topographic pattern (asymmetric bow tie with a skewed radial axis) (Figure 1). In "early" keratoconus, there are no slit-lamp findings, but scissoring is evident on retinoscopy with a dilated pupil. The typical topography (asymmetric bow tie with a skewed radial axis) is also present (Figure 2). Forme fruste keratoconus presents with no slit-lamp findings or scissoring on retinoscopy, but the typical topography (asymmetric bow tie with a skewed radial axis) is once again present (Figure 3). *Keratoconus suspect* is a catchall term to indicate a patient with inferior or central steepening on topography that the clinician suspects may progress to keratoconus<sup>1</sup> (Figure 4). The term is not synonymous with subclinical keratoconus, because the practitioner only knows it is subclinical once it has progressed to keratoconus. Many patients labeled as *keratoconus suspects* never develop clinical keratoconus.

## NORMAL CORNEAL TOPOGRAPHY

With a careful slit-lamp evaluation, dilated retinoscopy, pachymetry, and standard Placido-based videokeratography, surgeons can detect the vast majority of keratoconic subtypes preoperatively and thus prevent ectasia.<sup>1</sup> Despite many ophthalmologists' emphasis on posterior corneal curvature, I believe that the earliest signs of keratoconus appear anteriorly. Accurate diagnosis, however, demands a solid understanding of normal corneal topography, a category encompassing a large degree of variation.<sup>2,3</sup> In fact, using the absolute scale and sagittal topography, the classification scheme encompasses 10 patterns that all depict normal corneal topography (Figure 5).

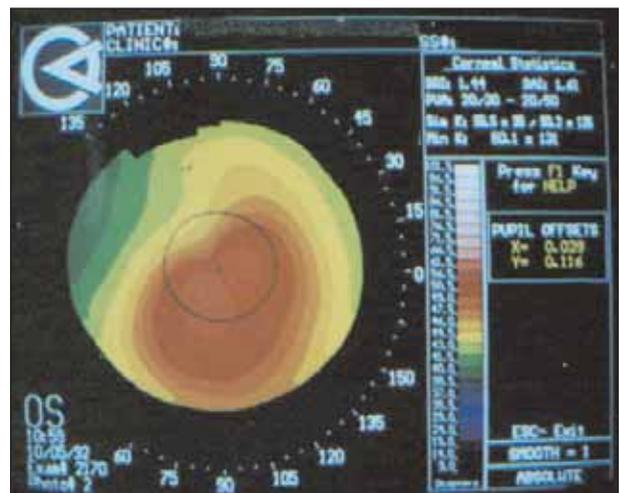


Figure 1. A keratoconic eye exhibits the typical topographic pattern of an asymmetric bow tie with a skewed radial axis.

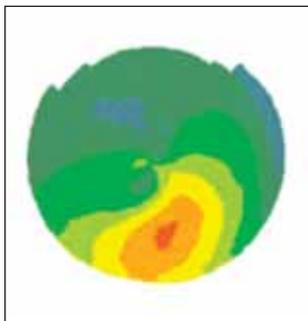


Figure 2. The same topographic pattern seen in Figure 1 is evident in this eye with early keratoconus.

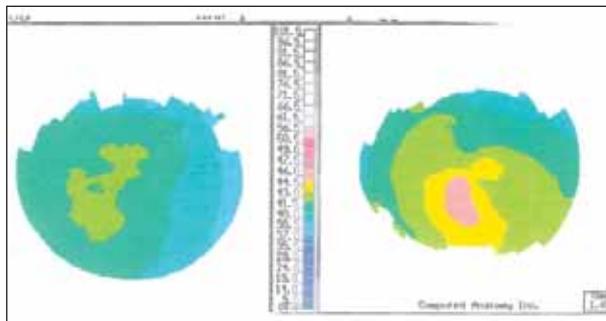


Figure 3. An eye with forme fruste keratoconus exhibits the same topographic pattern as the eyes shown in Figures 1 and 2.

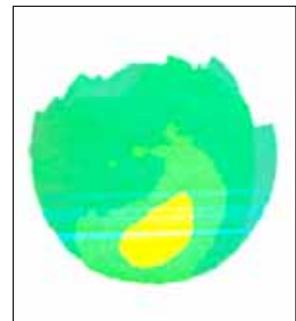


Figure 4. Inferior or central steepening on topography may progress to keratoconus.

Because normal corneas tend to be symmetrical within and between eyes, asymmetry is a clue to pathology. The pattern of an asymmetric bow tie with a skewed radial axis only occurs in 0.05% of the normal patient population, but it is almost universal in patients with keratoconus. Such individuals, even in the absence of clinical evidence of keratoconus, should be treated with a high degree of suspicion.<sup>4</sup>

## DETECTING PATHOLOGY

### Keratoconus

After ruling out contact lens-induced warpage, clinicians should look for the topographic pattern of an asymmetric bow tie with a skewed radial axis.<sup>5</sup> Indices such as the inferior/superior index that I developed<sup>6,7</sup> (Figure 6) are a useful adjunct. They are not diagnostic, however, and should be used only as an aid. Practitioners should take into account the entire clinical picture. Because there are several situations in which an abnormal inferior/superior value merely represents a false positive, it is not necessarily an indicator of a keratoconic subtype.<sup>8</sup> If the diagnosis on topography is still in doubt, the clinician should perform dilated cycloplegic retinoscopy in order to rule out early keratoconus.

Wavefront technology is an excellent adjunct to topography in the diagnosis of keratoconus, and I believe that future research will focus on using a combination of videokeratography and wavefront analysis to define keratoconic subtypes. A topographic map showing slight inferior steepening accompanied by significant coma on wavefront analysis, for

example, could be cause for great concern.<sup>9</sup> Preliminary research by my colleagues and me indicates that using a combination of the inferior/superior value (derived from topography) and vertical coma (derived from wavefront analysis) best separates early keratoconic subtypes from normals.<sup>10</sup>

### Pellucid Marginal Degeneration

I often serve as a defense witness in malpractice lawsuits for ectasia. Surprisingly, they most commonly involve cases of missed, early pellucid marginal degeneration versus missed early keratoconus. It seems that clinicians have become adept at diagnosing the latter but that they often overlook pellucid marginal degeneration, because it is rare. Most practitioners are aware that some inferior steepening on topography may be a warning sign, but early pellucid

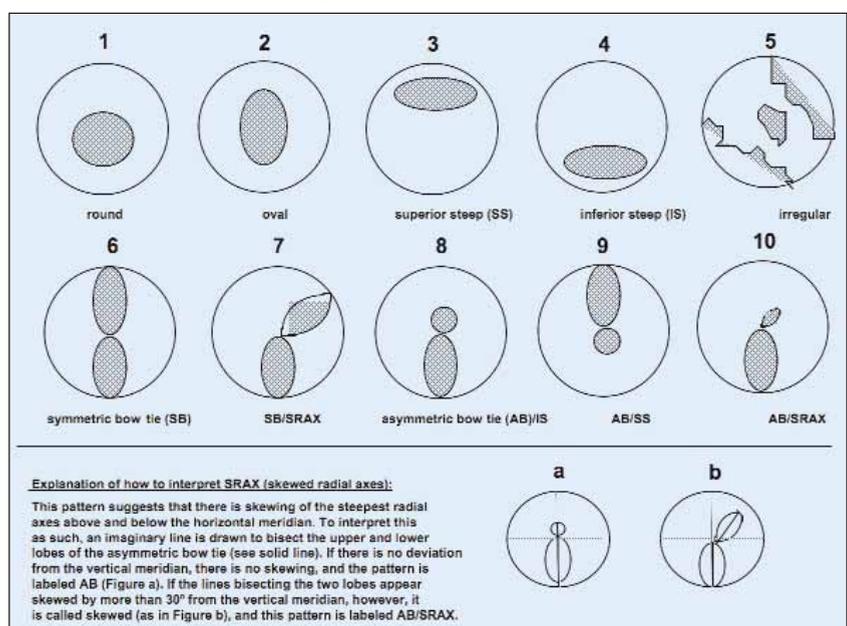


Figure 5. There is a spectrum of normal topography.

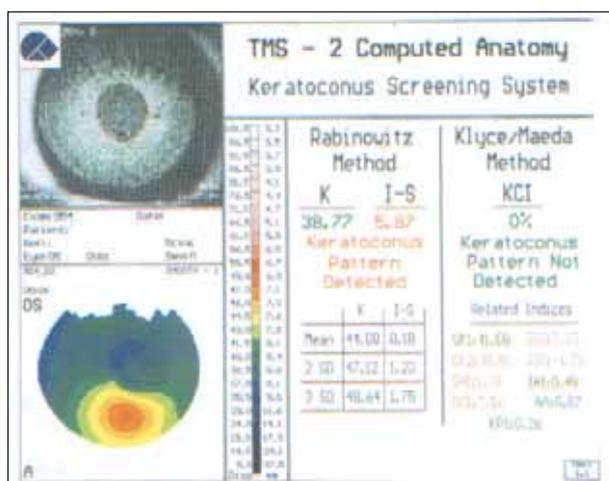


Figure 6. Indices can help in the diagnosis of keratoconus.

marginal degeneration presents as inferior flattening (Figure 7). It is in these cases that posterior topography may be relevant, but the keen-eyed clinician who is well trained in normal anterior topography will spot inferior flattening without having to rely on posterior-topography devices in most instances. In cases of obvious pellucid marginal degeneration, central flattening is accompanied by a crab-claw appearance on topography and a narrow band of corneal thinning from the 4- to the 8-o'clock position.

The take-home message is that clinicians must get used to the fact that inferior flattening is just as important as inferior steepening<sup>11</sup> (Figure 7).

### THE FUTURE?

In the future, a molecular genetic test for diagnosing early keratoconus and patients at risk for ectasia may become available that will be a Schirmer-like test using the polymerase chain reaction at the slit lamp. My colleagues at the National Eye Institute and I have demonstrated that aquaporin 5, a water transport gene, is suppressed early in keratoconus.<sup>12</sup> Although no mutations in this gene have been shown in keratoconus, the suppression of the gene might suffice to demonstrate the earliest stages of the disease. This test is still in development, however, and is not available for clinical use.

### CONCLUSION

Although a genetic test for keratoconus would be useful, it is currently only in the developmental stages. Surgeons already have several ways to identify keratoconus. The first step is to perform a careful slit-lamp examination. The second is to understand the variety present in normal corneal topography and to watch for asymmetry and identify abnormal topography. For eyes with suspicious topography, dilated retinoscopy can provide an important clue; if the

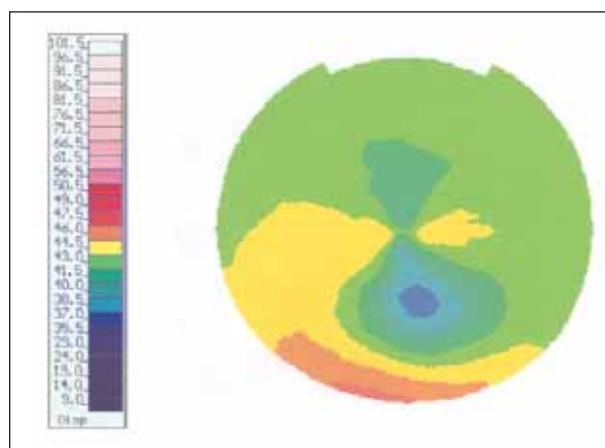


Figure 7. Early pellucid marginal degeneration presents as inferior flattening on topography.

findings are normal, contact lens-induced warpage may be the diagnosis and should be excluded. Indices and wavefront technology are also useful adjuncts. A combination of an abnormal inferior/superior value with high vertical coma on wavefront analysis should be treated as high risk. In addition, a family history of keratoconus is a significant risk factor. Clinicians should be sure to rule out pellucid marginal degeneration or suspected cases, because these patients may develop ectasia after LASIK as well. ■

Yaron S. Rabinowitz, MD, is a cornea and LASIK specialist. Dr. Rabinowitz is in private practice at Cornea Genetic Eye Institute in Los Angeles. He is also Director of Ophthalmology Research at Cedars-Sinai Medical Center and Director of Laser Eye Associates in Los Angeles. Dr. Rabinowitz may be reached at (310) 423-9640; [rabinowitz@cshs.org](mailto:rabinowitz@cshs.org).



- Rabinowitz YS. Keratoconus: update and new advances (major review). *Surv Ophthalmol*. 1998;42:297-319.
- Rabinowitz YS, Yang H, Elashoff J, et al. Videokeratography database of normal human corneas. *Br J Ophthalmol*. 1996;80:610-616.
- Rasheed K, Rabinowitz YS, Remba M, Remba D. Inter- and intra-observer reliability of a classification scheme for corneal topographic patterns. *Br J Ophthalmol*. 1998;82:1401-1406.
- Rabinowitz YS, McDonnell PJ. Computer-assisted corneal topography in keratoconus. *Refract Corneal Surg*. 1989;5:400-408.
- Rabinowitz YS. Corneal topography. *Curr Opin Ophthalmol*. 1993;4:68-74.
- Rabinowitz YS, Garbus JJ, McDonnell PJ. Computer-assisted corneal topography in family members of patients with keratoconus. *Arch Ophthalmol*. 1990;108:365-371.
- Rabinowitz YS. Videokeratography indices to aid in screening for keratoconus. *Arch Ophthalmol*. 1995;11:371-379.
- Rabinowitz YS, Wilson SE, Klyce SD, eds. *A Color Atlas of Corneal Topography: Interpreting Videokeratography*. New York, NY: Igaku-Shoin Medical Publishers; 1993.
- Rabinowitz YS. Ectasia after laser in situ keratomileusis. *Curr Opin Ophthalmol*. 2006;17:421-426.
- Jafri B, Li X, Rabinowitz YS. Wavefront analysis in the early detection of keratoconus. *Refract Corneal Surg*. In press.
- Rabinowitz YS. Pellucid marginal degeneration. An unsuspecting trap for the refractive surgeon. *Refractive Eye Care*. 2002 October.
- Rabinowitz YS, Dong L, Wistow G. Gene expression profile studies of the human keratoconus cornea for NEI Bank: a novel cornea-expressed gene and the absence of transcripts for aquaporin 5. *Invest Ophthalmol Vis Sci*. 2005;46:1239-1246.