# Could knowing your DNA help you take better care of your eyes?

Get the genetic test that gives you clear answers for optimal eye care

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# There's a chance you may have one of these genetic eye diseases

## Facts about keratoconus[1-6]

- In earlier studies, keratoconus was thought to affect as many as 1 in 1,800 people. Recent studies have shown that the prevalence could now be as high as 1 in 375, or even as high as 1 in 21 in certain populations[1,2]
- Common symptoms of keratoconus include blurred vision, seeing glare and halos around light, pain in the eye, and light sensitivity[3,4]
- Symptoms may show up in your teens or occur later in life[5]
- Keratoconus runs in families. If your parents or siblings have it, there's a good chance you may develop it[4,5]
- If you rub your eyes frequently, it may trigger your keratoconus or make it worse. Chronic inflammation to your corneas can also speed up the onset or progression of the disease[4,5]
- Eye rubbing, inflammation, or injuries to your cornea can trigger the progression of the keratoconus[6]

## Facts about corneal dystrophy[3,7-11]

- Around 1 in 1,100 people may have corneal dystrophy[7]
- Common symptoms include vision loss, eye pain, dry eye, sensitivity to light, glare, or the feeling that something is in your eye[8,9]
- Symptoms may be mild, moderate, or severe[3]
- Corneal dystrophy runs in families. If your parents or siblings have it, there's a good chance you will develop it
- Corneal dystrophy may show up in childhood or in your late 50s or 60s[3,10]
- To date, 70 different genetic mutations (TGFBIs) have been found to cause corneal dystrophy[11]

# Learn about the AvaGen DNA eye test

Genetic tests like **AvaGen** are the future of medicine and help doctors offer more personalized care for patients.

## Test highlights

- Utilizes a custom panel
- Examines more than 1,000 variants across 75 genes for keratoconus and more than 70 TGFBI mutations for corneal dystrophy
- Tells you if you are likely to develop keratoconus before symptoms even appear
- Uses sophisticated algorithms to detect very rare variants, which is key to understanding complex eye diseases such as keratoconus and corneal dystrophy
- Helps you take charge of your eye health by providing you with answers much earlier than ever before, enabling you to make healthy lifestyle decisions

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# There are good reasons to get tested for keratoconus and corneal dystrophy

1. You want to take positive steps towards maintaining your eye health today and in the future.
2. Early diagnosis and awareness of risk factors will give you the opportunity to make smart decisions for your long-term vision.
3. A family member has keratoconus or corneal dystrophy, which means your risk of developing either disease is greatly increased.
4. You are in the process of making vision correction decisions for yourself or other family members.

## You should get tested if:

- You are considering refractive surgery
- You have suspicious gray or white spots (protein deposits) on your corneas
- You have a family history of corneal spots, corneal dystrophy, vision changes, or a family member who had a corneal transplant for unknown reasons
- You have a family history of keratoconus

If you said **"yes"** to any of the above, ask about **AvaGen**, the genetic DNA test for keratoconus and TGFBI corneal dystrophy.

## Here's what you should know about the AvaGen genetic test

- Testing is non-invasive
- A sample is taken from the inside of your cheek with a swab
- AvaGen can tell you if your DNA shows genetic variants or mutations associated with keratoconus or corneal dystrophy
- AvaGen may tell you that you are at risk for keratoconus and/or give you a definite **"yes" or "no" answer** to whether you have corneal dystrophy
- You may request to have your DNA stored in your medical records or stored in our secure database and made available to you for other health conditions in the future

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# It's quick and easy to get tested

1

## REQUEST:

Your eyecare professional will collect the sample in the clinic, or they will order the **AvaGen** test and inform you when it arrives

2

## SAMPLE:

Your eyecare professional will swab the inside of your cheek

3

## LAB:

Your eyecare professional will send your sample to the lab

4

## RESULTS:

Your eyecare professional will discuss the test results and next steps with you

5

## COUNSELING:

You will be connected with a network of genetic counselors if your test results come back positive. A genetic counselor can help explain your results, what they mean for your health, and what to expect next

Ask your eyecare professional for a referral for a genetic counselor.

# About Avellino, makers of AvaGen

## Who is Avellino?

- A global precision medicine leader with its headquarters in the United States and operations worldwide
- Empowering eyecare professionals by giving them the data they need to make better decisions for their patients
- Driven to help patients lead healthier lives
- Pioneering first-in-class genetic tests and therapies for corneal dystrophies, keratoconus, and other conditions

## First and only genetic test for keratoconus and TGFBI corneal dystrophy

Avellino Labs is regulated under the Clinical Laboratory Improvement Amendments (CLIA) of 1988 and is licensed by the United States Division of Laboratory Services under the Center for Clinical Standards and Quality.

Avellino Labs is HIPAA compliant.

**References: 1.** Torres Netto EA, Al-Otaibi WM, Hafezi NL, et al. Prevalence of keratoconus in paediatric patients in Riyadh, Saudi Arabia. *Br J Ophthalmol.* 2018;102(10):1436-1441. **2.** Godefrooij DA, de Wit GA, Uiterwaal CS, et al. Age-specific incidence and prevalence of keratoconus: a nationwide registration study. *Am J Ophthalmol.* 2017;175:169-172. **3.** American Academy of Ophthalmology website. https://www.aao.org/eye-health/diseases/a-z. Accessed August 8, 2019. **4.** Johns Hopkins Medicine. Keratoconus. https://www.hopkinsmedicine.org/health/conditions-and-diseases/keratoconus. Accessed August 6, 2019. **5.** Gordon-Shaag A, Millodot M, Shneor E, Liu Y. The genetic and environmental factors for keratoconus. *Biomed Res Int.* 2015;2015:795738. **6.** Ionescu C, Corbu C, Tanase C, et al. Inflammatory biomarkers profile as microenvironmental expression in keratoconus. *Dis Markers.* 2016;1243819. **7.** Musch DC, Niziol LM, Stein JD, Kamyar RM, Sugar A. Prevalence of corneal dystrophies in the United States: estimates from claims data. *Invest Ophthalmol Vis Sci.* 2011;52(9):6959-6963. **8.** Bourges JL. Corneal dystrophies. *J Fr Ophtalmol.* 2017;40(6):e177-e192. **9.** National Institutes of Health website, TGFBI gene. https://nei.nih.gov/health/cornealdisease. Accessed August 18, 2019. **10.** Mayo Clinic. Fuchs' dystrophy. https://www.mayoclinic.org/diseases-conditions/fuchs-dystrophy/symptoms-causes/syc-20352727. Accessed August 23, 2019. **11.** Chao-Shern C, DeDionisio LA, Jang JH, et al. Evaluation of TGFBI corneal dystrophy and molecular diagnostic testing. *Eye (Lond).* 2019;33:874-881.**12.** Cope JR, Collier SA, Nethercut H, Jones JM, Yates K, Yonder JS. Risk behaviors for contact lens-related rye infections among adults and adolescents, United States, 2016. *MMWR Morb Mortal Wkly Rep.* 2017;66:841-845.

# See if you are a good candidate for the AvaGen DNA eye test

- I want vision correction surgery
- I would like contact lenses[12]
- My vision has continuously gotten worse over the last several years
- Someone in my family has keratoconus
- Someone in my family has corneal dystrophy or corneal spots
- I was told I have unusual spots (protein deposits) on my corneas
- Someone in my family has had a corneal transplant
- I don't know the genetic history of either of my parents
- I have never had a corneal wound or penetrating eye injury

Be sure to tell your physician if you have **checked 1 or more** of these boxes.

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1505 Adams Drive, Suite B2, Menlo Park, CA 94025
infousa@avellino.com | (650) 396-3741 | Visit avellino.com/patients