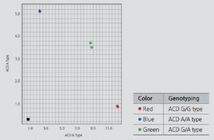
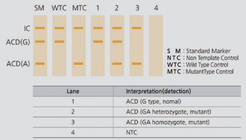
Product and Ordering Information

Our Real-Time and Conventional Genotyping Kits: A Comparison

	DiaPlexQ™ Genotyping Kit (Real-Time)	DiaPlexC™ Genotyping Kit (Conventional)
Detection Method	Real-Time PCR	Conventional PCR
Specimen	Blood, Oral Epithelial Cell, Hair Root	Blood, Oral Epithelial Cell, Hair Root
Compatible Instrument	ABI 7300, 7500, 7500HT Fast, 7900HT Fast Qiagen Rotor Gene Q Roche LightCycler 480, 1536 Bio Rad CFX96™, Agilent Technology Mx qPCR	PCR Thermal Cycler
Inspection Time	1 hour and 40 minutes	2 hours and 10 minutes
Certificate	CE IVD	CE IVD
Result Analysis	Allelic discrimination plot, Amplification plot  ► *DiaPlexQ™ ACD Genotyping kit*	Agarose gel  ► *DiaPlexC™ ACD Genotyping Kit*

Lane	Interpretation(detection)
1	ACD (G type, nomal)
2	ACD (GA heterozygote, mutant)
3	ACD (GA homozygote, mutant)
4	NTC

Ordering Information

Cat. No.	Product
SQH26-K100 (100 tests) SQH26-K020 (20 tests)	DiaPlexQ™ Avellino Corneal Dystrophy (ACD) Genotyping Kit
SHG06-K100 (100 tests) SHG06-K020 (20 tests)	DiaPlexC™ Avellino Corneal Dystrophy (ACD) Genotyping Kit

SolGent Co., Ltd.
Solutions for Genetic Technologies
www.solgent.com
global@solgent.com Tel: +82 (0)31-608-5693

SolGent

Avellino Corneal Dystrophy

The DiaPlexC™/Q™ ACD Genotyping Kit

SolGent
Solutions for Genetic Technologies

Headquarters
3F, 32 Techno 6-ro, Yuseong-gu, Daejeon, 305-509, Korea Tel) +82-42-863-5685 Fax) +82-42-936-5695

Overseas Business Department
701-3, Pangyo Innovalley Bldg. B, Pangyo-ro 253, Bundang-gu, Seongnam-si, Gyeonggi-do, 463-400, Korea
Tel) +82-31-608-5693 Fax) +82-31-608-5694

CE IVD

What is Avellino Corneal Dystrophy?

Avellino corneal dystrophy (ACD), also commonly known as Granular Corneal Dystrophy Type 2 (GCD2), is one of the autosomal-dominant dystrophies among the granular (Groenouw Type 1), Lattice Type 1 and Reis-Bucklers.
ACD is a transforming growth factor beta-induced (TGFBI) gene, formally known as BIGH3 codon 124(R124H) associated with the development of corneal dystrophies. It is an inherited disease more severe in homozygous than heterozygous patients.[1-3]

The course of the dystrophy can vary with each person, although there are two common patterns: In some cases, symptoms can present in early life, leading to blindness before the age of 10.
Another common trait is for ACD to appear gradually throughout a lifetime.[3-5]
ACD is NOT curable, thus prevention of exacerbation is the only method available for those with the dystrophy.[4-6]
It is vital to identify ACD in patients in order to correctly advise on the future course of the dystrophy, and on which aspects in their lives may have a detrimental affect on the condition of their eyesight.
For example, LASIK surgery will cause symptoms to rapidly accelerate, even if none were visible through manual inspection prior to surgery.[3-8]

Persons regarded as high risk:
- People with a family history of the disease
- Those considering LASIK, LASEK or PTK
- People living or working in high UV environments

Despite the danger associated to the quality of life for sufferers with ACD, with correct care ACD can be managed without the potentially extreme symptoms presenting throughout a patients life.. For this to be a possibility, patients and their doctors need to know they have the disease so that fully informed decisions can be made regarding treatment for the patients eyes, as well as everyday care and management.

It is our aim to provide accurate and reliable detection of ACD in patients through a genetic test, enabling awareness and allowing the patient to avoid mitigating factors that would have caused unnecessary exacerbation of the dystrophy.

Avellino Corneal Dystrophy Throughout The World

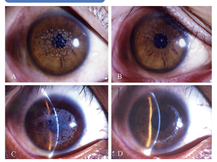
An increase in media coverage is bringing ACD to the attention of people considering LASIK and LASEK is reflected in the increase of reported cases. Furthermore, patients previously misdiagnosed as having granular corneal dystrophy are now being correctly diagnosed as suffering from Avellino Corneal Dystrophy[1]
As knowledge and awareness of ACD has developed, some opticians in China, Japan and Korea now refuse access to LASIK / LASEK in patients with ACD because of the harmful effects the procedure has on patients..

Countries	Frequency of ACD among ALL Corneal Dystrophies	References
Korea	90%	Lee et al. Ophthalmic Epidemiol. 2010 June;17(3)
Japan	70%	Shigeo et al. Jpn J Ophthalmology 46, 469-471(2002)
China	80%	Juhua Yang et al. Molecular Vision 2010; 16:1186-1193
India	60%	Arch Ophthalmol. 2009;127(10):1373-1376
Germany	60%	Lang et al. Cornea 1987;6(3):209-11
Canada	65%	Elvira et al. Arq Brass Oftalmol. 2012;75(6):390-3

Case Studies

Environmental situations can cause the effects of ACD to worsen, such as working or living in high UV environments.
The symptoms of ACD drastically accelerate following LASIK, LASEK and PTK surgery [5-9]

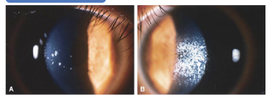
case 1



Recurrence location after excimer laser phototherapeutic keratectomy (PTK) for corneal dystrophy. References 6

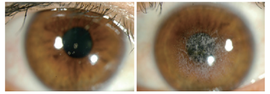
A, Granular dystrophy with breadcrumb-like corneal opacity.
B, Early recurrence 18 months after PTK for granular dystrophy.
C, Macular corneal dystrophy with thick corneal opacity.
D, Recurrence circling the corneal periphery 12 months after PTK.

case 2



A, Discrete granular opacities in the anterior stroma of the un-operated eye (right eye) 5 years after LASIK in the opposite eye.
B, Numerous dense, confluent granular opacities are seen centrally in the interface between the flap and residual stromal bed of the operated eye (left eye)5 years after LASIK. References 7

case 3



Slit-lamp photograph of granular deposits of anterior stroma in Avellino corneal dystrophy. The right eye shows discrete stromal opacities with intervening clear stroma and sparing of the periphery.
The left eye shows confluent and ground-glass like opacities sparing of the periphery. References 8

case 4

Dystrophy Patients

Mutations		No. of Cases	No. of Families	Disease*
R124H	Heterozygous	39	38	ACD
R124C	Heterozygous	2	2	LCD
R555W	Heterozygous	4	4	GCD
Total		45	44	

*ACD: Avellino corneal dystrophy, LCD: Lattice corneal dystrophy, GCD: Granular corneal dystrophy.

References 9

References

1 Kim, J.W., Kim, H.M. and Song, J.S., Journal of the Korean Ophthalmological Society 01/2008; 49(9). DOI: 10.3341/jkos.2008.49.9.1431
2 Klintworth GK. Advances in the molecular genetics of corneal dystrophies. Am J Ophthalmology 1999; 128: 747-54.
3 Konishi M, Mashima Y, Nakamura Y, et al. Granular-lattice (Avellino) corneal dystrophy in Japanese patients. Cornea 1997; 16: 635-8.
4 Kocak-Atlintas AG, Kocak-Midillioglu I, Akarsu AN, Duman S. βigh gene analysis in the different diagnosis of corneal dystrophies. Cornea 2001; 20: 64-8.
5 Weiss 2008, Roo 2004, Aldave 2007, Lee 2008, Kim 2008
6 Min Chen & Lixin Xie. Features of Recurrence after Excimer Laser Phototherapeutic Keratectomy for Anterior Corneal Pathologies in North China. Ophthalmology June 2013, 1179-1185
7 Jun et al, Avellino Corneal Dystrophy after LASIK Ophthalmology Volume 111, Number 3, March 2004 doi:10.1016/j.ophtha.2003.06.026
8 Na KS, Kim MS (2011) An Unusual Form of Avellino Dystrophy after Laser in situ keratomileusis: A Late Onset or Recurrence?.J Clinic Experiment Ophthalmol 2:172. doi:10.4172/2155-9570.1000172
9 Yoshida et al., Jpn J Ophthalmology 2002, 46, 469-471

Benefits for the Doctor and Patient

The current manual method used to identify ACD in patients is unreliable, and cannot show the presence of ACD until symptoms are showing. The effects of ACD will worsen after LASIK / LASEK regardless of whether there were symptoms present before surgery.
Misdiagnosis will cause many problems for the patient during the course of their life. It is therefore vital to reliably identify patients with ACD to avoid further damage.

A genetic test is the first step towards preventing the dystrophy from worsening and really starting to affect the quality of life for the individual.

A pre-surgery genetic test is an essential qualifying test for patients waiting for LASIK / LASEK / PTK, allowing for a fully informed decision to be made by each of the parties involved upon its result. Doctors will be able to proceed with confidence knowing the surgery will not result in eventually poorer eyesight for their patients, helping them to make the best decision. Furthermore, genetic testing for ACD is yet to report cases of misdiagnosis, supporting the reliability of this method.

Patient	Doctor
Eye safety	Avoid misdiagnosis.
Prevention	Proceed with confidence.
Confirm the safety of future generations.	Competitive advantage.

Test Process

Sample Collection → Delivery → Testing and Result Analysis → Consultation