The Value of Reliable Genetic Testing in Refractive Surgery Candidates

To the Editor:

The corneal deposits seen in heterozygote granular corneal dystrophy type 2 (GCD2) can be observed in individuals with variable appearances. When the deposits are small and at times undetectable, only genetic analysis is effective for the confirmative diagnosis. Many refractive surgeons have adopted genetic testing prior to refractive surgery to verify that their patients are free from the risk of exacerbation of GCD2 following LASIK, LASEK, or photorefractive keratectomy.

As an example, a 30-year-old Korean woman visited the Corneal Dystrophy Clinic of Severance Hospital, Seoul, Korea in April 2016 with decreased visual acuity and fine central corneal deposits in both eyes (Figures 1A-1B). The corneal findings were representative of the typical appearance of exacerbated GCD2 after LASEK, as previously reported. She had received uneventful bilateral LASEK in September 2009 at a local Korean clinic. On presentation, her corrected distance visual acuity was 20/32 in the right eye and 20/63 in the left eye. Preoperative manifest refraction was -4.50 -2.50 × 175 in the right eye and -6.75 -1.50 × 175 in the left eye, yielding 20/20 in both eyes. One year after LASEK, the patient began to notice decreased visual acuity.

Her preoperative medical record from the local clinic described four small subepithelial opacities on her right cornea with no deposits on the left cornea (Figure 1C). A preoperative gene test report for GCD2 from a local commercial company for detecting GCD2 mutation reported the patient as negative for the mutation. This company used a patented adhesive tape technique, applying the tape to the forearm for 20 seconds and then peeling it off to obtain the epidermal keratinocytes for DNA extraction, followed by polymerase chain reaction (PCR) and DNA sequencing. The additional collection of three hair follicles was used to ensure sufficient quantity of DNA. The company offered GCD2 gene testing services in 2009 and subsequently stopped testing in 2010. The surgeon performed LASEK after receipt of the negative GCD2 report.

The patient was retested for GCD2 mutation after informed consent in our hospital using PCR and sequencing of DNA from peripheral blood with two different primers, which identified the patient to be a GCD2 heterozygote. In addition, the Avellino Universal Test (performed by Avellino Lab Korea using Avellino Lab USA’s proprietary technology), which uses buccal epithelial cells collected with a cotton swab, reported the patient to be a GCD2 heterozygote. These new tests demonstrate that the genetic test system performed in 2009 was inaccurate at notifying the surgeon of the patient’s GCD2 condition. The patient was not able to confirm whether she provided the three additional hair follicles for the test performed in 2009, which may have contributed to the test error.

This case report shows the importance of reliable genetic testing before certain refractive surgical procedures. Tests performed by genetic testing laboratories using methods validated with clinical trials and the Clinical Laboratory Improvement Amendments certification or equivalent validation would provide the refractive surgeon with high confidence in the reported test results. Genetic testing that can detect all TGFBI mutations would certainly be welcome in the future.

REFERENCES


Figure 1. Slit-lamp photographs of the (A) right eye and (B) left eye show numerous fine white granular deposits in the central zone 6 years after bilateral LASEK. (C) The redrawing of the preoperative medical record of the local clinic shows four small subepithelial opacities only in the right cornea.


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