Twin Study Implicates Genetic Factors in Dry Eye Disease

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Dry eye is a common chronic ocular condition that significantly reduces the quality of life of afflicted individuals. As well as a frequent cause of ocular irritation, dry eye can lead to visual disability, and may compromise the results of corneal, cataract, and refractive surgery.¹ The average cost to US society of managing dry eye has been estimated at $55.4 billion.²

Dry eye disease encompasses a group of disorders of the tear film; attributed to either reduced tear production or excessive tear evaporation, or both. There may be inconsistent correlation between symptoms and signs, making clinical diagnosis problematic. Current treatments are largely palliative. Although symptoms can improve with treatment, there is no cure.³

Epidemiologic information on dry eye disease has been limited by the variety of definitions employed and the lack of diagnostic tests of high sensitivity and specificity.⁴ Female sex, increasing age, Sjögren’s syndrome, and smoking have been identified as risk factors; other systemic diseases are also associated. Understanding the genetic factors of a disease can provide insights into the pathogenesis of disease, and hence lead to more effective treatment. To date, there is very little information on the role genetics plays in the dry eye phenotype, or the relative importance they have to environmental factors.

Vehof et al.⁵ have employed an elegant classical twin study using a cohort of British, middle aged, elderly female twins. Despite the difficulties in phenotyping this multifactorial disease, they have demonstrated that genes are moderately important in dry eye disease. A heritability of approximately 30% was indicated for symptoms and 40% for diagnosis, with a varying heritability of 25% to 80% for various selected signs; the unique environment explained the remainder of the variance. Vehof et al.⁵ have paved the way for further genetic studies by showing that genetic factors play a significant role in dry eye disease.

References