What Is New in Ophthalmic Research – Focus on Genetics

Higher-Order Aberrations Help in Understanding the Etiology of Myopia

Dirani et al., Melbourne, Vic., Australia

Ophthalmic Res 2009;41:142–147

Myopia is a complex eye condition, with both environmental and genetic factors contributing to its developments. There have been several large twin studies that have collectively found a genetic basis to myopia; however, these studies have typically focused on low-order aberrations. The authors resort to quantify the genetic component to higher-order aberrations that are typically not routinely assessed in ophthalmic clinics, even though they make up a part of one's refractive state. They examined the aberrations (low and high) of a subgroup of twins who participated in the Genes in Myopia twin study, and found evidence to support a genetic role for higher-order aberrations. Therefore, they believe that it is essential to consider higher-order aberrations in future myopia research, in particular those concerned with its etiology.

Cerulean Cataract in a Chinese Family Is Associated with a Gene Conversion Mutation in β -Crystallin B2

Wang et al., Harbing, China

Ophthalmic Res 2009;41:148–153

The authors demonstrate a mutation (NM_000496.2: c.463C→T, p.Q155X) in the *CRYBB2* gene locus associated with congenital cerulean cataract in a Chinese family. In addition, they found another transition (NM_000496.2: c.471C→T). These two transi-

tions are identical to the sequence of *CRYBB2P1*. It is suggested that the gene conversion between *CRYBB2* and *CRYBB2P1* could be the developmental mechanism of congenital cerulean cataract. These findings provide another evidence for β -crystallin B2 associated with cerulean cataract. Future studies may address the detailed mechanism of cataract formation and genotype-phenotype correlation.

Birth Weight Does Not Predict Myopia Development in Twins

Dirani et al., Melbourne, Vic., Australia

Ophthalmic Res 2009;41:154–159

Understanding the complexities of myopia has taken the frontline in ophthalmic research, with particular focus on its development. There is a substantial amount of evidence to support a genetic and environmental role in myopia; however, a conclusive etiology is yet to be defined. The authors sought to examine whether birth weight was related to the development of myopia. They assessed over 1,200 twins and found no significant associations between low birth weight and the development of myopia. Even though their results are negative, it has helped them pave the way for future research into myopia. They believe that more focus is needed in identifying the gene(s) associated with myopia and how they may be interacting with already identified environmental factors. There is also a need to explore epigenetic factors that may explain a large part of the differences of myopia prevalence observed among varying populations.

Uwe Pleyer, Editor-in-Chief