Predicting Children At-risk of Myopia

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. Genetic Testing Using Saliva Sample .

Myopia (short-sightedness) is an eye disorder that develops during school-age, and which is usually correctable using spectacles or contact lenses. Currently, myopia affects about 80% of Hong Kong children by the time children reach 18 years-old. In extreme cases, “high degree” myopia can cause irreversible blindness – indeed, it is rapidly becoming the most frequent cause of untreatable blindness in south east Asia.

New optical and pharmacological treatments for slowing myopia progression are being developed, however determining which children are at high risk is imprecise: the best current (non-genetic) method has sensitivity 62.5% and specificity 81.9%. Due to the high heritability of myopia (50-80%) genetic testing has the potential to improve the detection of at-risk children at an early age. Treating these at-risk individuals will reduce their risk of blindness in later life.

Published genome-wide association studies (GWAS) for myopia have identified 39 common genetic variants that predict at-risk subjects (explaining approximately 10% of the variance in “refractive error”). Larger-scale GWAS studies are expected to increase the sensitivity and specificity with which at-risk children can be detected.

Representative Publication