THE HONG KONG POLYTECHNIC UNIVERSITY
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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.

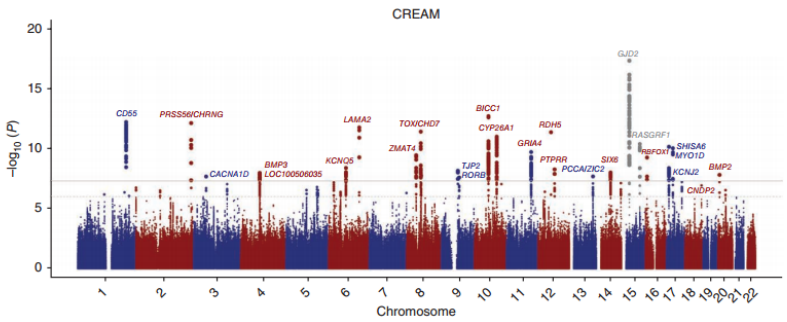

Figure 1 Manhattan plot of the GWAS meta-analysis for refractive error in the combined analysis ($n = 45{,}758$). The plot shows $-\log_{10}$-transformed P values for all SNPs. The upper horizontal line represents the genome-wide significance threshold of $P < 5.0 \times 10^{-8}$; the lower line indicates P value of 1×10^{-5}. Previously reported genes are shown in gray. The *RBFOX1* gene is also known as *A2BP1*.

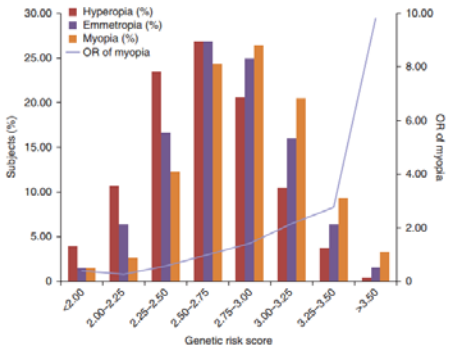

Figure 2 Genetic risk score for myopia. Distribution of subjects from Rotterdam Study 1–3 ($n = 9{,}307$) with myopia (SE ≤ −3 diopters (D)), emmetropia (SE ≥ −1.5 D and ≤ 1.5 D) and hyperopia (SE ≥ 3 D) as a function of the genetic risk score. This score is based on the regression coefficients and allele dosages of the associated SNPs for all 26 loci identified in the meta-analysis. Mean OR of myopia was calculated per risk category, using the middle risk score category (risk score of 2.50–2.75) as a reference.

Representative Publication

Verhoeven VJM, et al. Genome-wide meta-analyses of multiancestry cohorts identify multiple new susceptibility loci for refractive error and myopia. Nature Genetics 2013;45:314-318.

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