

Genetic study of IgA nephropathy in China

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IgA nephropathy (IgAN) is the most common primary glomerulonephritis among patients undergoing renal biopsy; several lines of evidence support the importance of genetic risk factors to the pathogenesis of IgAN.

Our previous GWAS in Chinese Han population for IgAN have identified associations at chromosome loci 17p13 and 8p23. In addition, we found multiple associations in the *MHC* region and confirmed a previously reported association at 22q12. For the post-GWAS study, we will do the further study to localize the susceptibility genes associated with IgAN. We has done the data mining of IgAN GWAS data, discovered novel three susceptibility loci, validated a recently reported association on 16p11.2, and identified three independent signals within the *DEFA-DEFB* locus at genome-wide significance. The effect of gene-gene interaction on the susceptibility and clinical manifestations of IgAN has been analysis by using multifactor dimensionality reduction (MDR) method. The result showed a potential gene-gene interaction on the susceptibility of IgAN, and gene-gene interaction had combined effects on macroscopic hematuria and the formation of crescent in IgAN patients. The study of polymorphism of *DEFA* genes discovered seven SNPs within *DEFA* genes were significantly associated with IgAN. Functional study suggested that polymorphisms within *DEFA* genes are involved in gene transcriptional regulation. All of our work support the evidence of the genetic polymorphisms can influence the susceptibility to IgAN.