



DR KHOR CHIEA CHUEN

Assistant Professor
Department of Biochemistry, NUS Medicine
Saw Swee Hock School of Public Health
Group Leader & Senior Principal Investigator
Genome Institute of Singapore (GIS), A*STAR

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Genetic susceptibility to common diseases. How far we have come, and how much more there is to do

In the last few years, genome-wide association studies (GWAS) have revolutionized the field of genetics and heredity dramatically. Starting from twenty years of false positive, irreproducible results from candidate gene selections, the GWAS has allowed a totally unbiased view into human disease associations, thus allowing genuine and unexpected genetic hits which are robustly replicable. Nonetheless, critics of the GWAS have repeatedly pointed out that

- a) Genetic associations are not causations
- b) These whole genome genetic studies are often meaningless due to the small effect sizes.
- c) There is little medical relevance apart in GWAS studies.

Here, I will discuss with you what we have done, and what we are planning to do in terms of follow-up experiments. Most of the follow-up experiments will involve deep re-sequencing of some sort. In the end, I feel that one of the reconciling features between genetic association studies with human biology / disease is that human diseases are often a consequence of mutations in our genetic code (DNA), which should then be visible either at the RNA and protein end, and ultimately impact function.



Department of Biochemistry
Yong Loo Lin School of Medicine
National University of Singapore
8 Medical Drive, MD 7, #02-08, Singapore 117597



+65 6516 3682



bchsec@nus.edu.sg
medicine.nus.edu.sg



Biochemistry.NUS



Department of Biochemistry
Yong Loo Lin School of Medicine