DEPARTMENT OF BIOCHEMISTRY

SEMINAR



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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.



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