

Institute of Aerospace Medicine

Institute Seminar, June 13, 2017, *Abstract*

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Big data in medical research

Metabolic diseases like obesity or type 2 diabetes are common complex diseases with several different environmental as well as genetic factors involved. This lecture gives a short overview of the genetic characteristics of metabolic diseases. Moreover connections between metabolite-gene associations and their role in metabolic diseases will be presented.

Serum metabolite concentrations allow a direct readout of biological processes and association of specific metabolomic signatures with complex diseases. These metabolites can rather easily be measured in large biobanks. Genome-wide association studies (GWAS) have identified many risk loci for complex diseases, but effect sizes are typically small and information on the underlying biological processes is often lacking. Associations with metabolic traits as functional intermediates can overcome these problems and potentially inform individualized therapy. We identified different genetic loci associated with blood metabolite concentrations, of which many exhibit effect sizes that are unusually high for GWAS and account for 10-60% of metabolite levels per allele copy. Our associations provide new functional insights for many disease-related associations that have been reported in previous studies, including cardiovascular and kidney disorders, type 2 diabetes and obesity. Our studies advance our knowledge of the genetic basis of metabolic individuality in humans and generate many new hypotheses for biomedical and pharmaceutical research.