“Towards using Genetic Information in Health Care and Prevention”

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Most -if not all- human diseases and their risk factors have a genetic component, implying that variance among individuals in susceptibility, treatment response and/or progression, is determined -in part- by genetic variation. DNA analysis technology is developing continuously and allows sequencing a human genome in <24hours (expensive) but also analyzing millions of SNPs in millions of DNA samples using arrays (cheap). Human genome sequencing projects, such as the European 1 million Genomes (1MG) Project, have uncovered hundreds of millions of such genetic variants, but it has been array/chip technology -applied in cohort studies and biobanks- that has identified tens of thousands of genetic factors for common diseases by Genome Wide Association Studies (GWAS). Altogether, this has led to genetic information now entering the hospital clinic in a broad sense, whereby –in theory- all patients can be assessed for (clinically actionable) DNA mutations and polygenic risk scores, next to pharmacogenomics information and their ancestry, blood groups and HLA profiles, for example. Such genetic information can help clinicians in decision making for diagnosis and treatment, and to provide self-empowerment for patients for prevention. Such a program exploring these opportunities, called GOALL (Genotyping on ALL patients) is running at Erasmus MC in Rotterdam, The Netherlands. However, also outside of the (academic) hospitals, applications of using genetic information are explored, such as in population screening programs, e.g., for breast cancer or Familial Hypercholesterolemia. I will describe aspects of these developments, highlight examples, and provide an outlook on the future.