

From Microarrays to Next Generation Sequencing: The impact of genomics on modern medicine

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In the last decade Illumina developed sequencing by synthesis (SBS) as the leading technology for next generation sequencing that is today providing over 90% of all DNA sequencing data globally. From targeted sequencing of pre-enriched marker regions over metagenomics studies on microbial communities to whole exome and whole genome sequencing of patients in the clinical routine and entire populations – NGS technology is today present not only in all major clinical research centers but also in routine clinical praxis. Rare diseases, that until recently took years to be diagnosed, can now be identified much more rapidly and compared with similar cases in global databases. This dramatically shortens the long and erratic path to diagnose these rare disorders, relieve patients and their families and can sometimes even indicate a promising treatment strategy. NGS-based tests can not only help to evaluate pre-dispositions for inheritable cardiovascular diseases or cancer, it also is now used to develop non-invasive tests for the early detection of cancer and to predict the chances for treatment success. The same principle is used to sequence fetal DNA in the bloodstream of the mother, which allowed to develop routine assays for non-invasive prenatal testing (NIPT) for embryonic aneuploidy, which can be the cause of Down Syndrome in the case of trisomy-21.