



SNP ANALYSIS IN HUMAN GENOME

Genetic variations strongly influence all sorts of diseases and the peculiarity of certain phenotypes. This is important for diagnosis of diseases, compatibility of certain drugs, and determination of potential personal disease risk. Furthermore, such procedures make it necessary to examine potentially pathogenic variants in appropriate cohorts.

Most frequent variations are single base exchanges, so called SNPs (single nucleotide polymorphisms). Apart from SNPs, variations in the number of copies of certain genomic regions (CNPs, copy number polymorphisms) are important too.

Genotyping technology GENOSNIP is surpassingly suitable to analyze almost all SNPs and most CNPs. It is based on single base extension and MALDI-TOF. Several SNPs (up to 10) can be measured with one reaction, resulting in a decreased need of sample material per analysis.

Unique Feature

There is an option for automated analysis in larger projects. The technology is easily applicable on a wide range of research topics where SNP- and CNP variations either play a role or are supposed to do so.

Methods

DNA is extracted from tissue, saliva or blood. In larger studies with some hundred persons and many genetic markers, DNA can be aliquoted by a laboratory robot. Assay design and assay validation are performed. Genotyping is done using PCR and single base extension with subsequent mass spectrometric analysis. Again, large scale studies are supported by automation. Spectre analysis and statistics are done with appropriately optimized software.

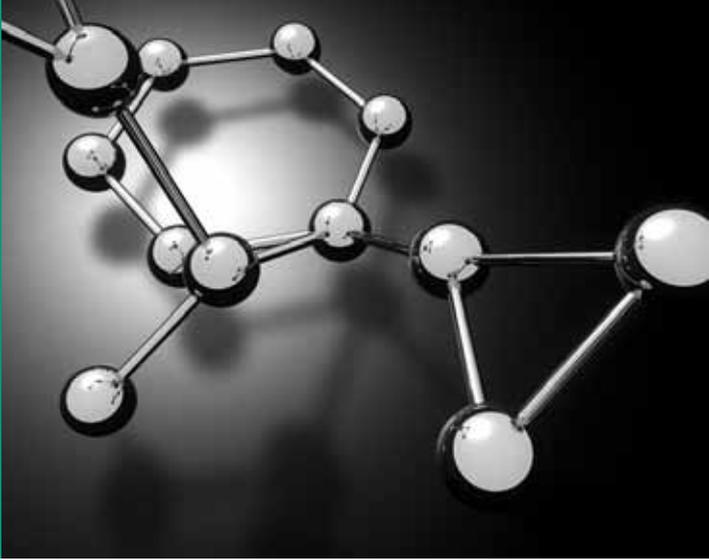
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Investigation Methods

- DNA extraction
- DNA aliquoting
- Assay design
- DNA amplification and genotyping
- Statistical analysis

Selected Applications

- Diagnosis of genetic diseases
- Epidemiological analysis of supposed genetic risk factors
- Development of optimized therapies (“personalized medicine”)
- Determination of genetic risk for certain diseases

Reference Project

Almost all diseases are a result of the combined influences of external environmental factors and internal genetic factors. The relation between these factor types that lead to a disease is surprisingly static. Even for dyslexia, a reading and writing disorder, which was tragically misinterpreted as a lack of intelligence, a genetic background could be found. But this background does not influence intelligence. Due to this reason specific teaching and learning techniques can help affected children, if they are diagnosed early enough. Those techniques are effective especially when they are applied long before the diagnostic window of current performance tests: long before school. Our aim is to identify mutations in single base pairs (SNPs) in the human genome that correlate with dyslexia. With this knowledge it would be possible to estimate the individual risk very early. And with early therapies in pre-school age later difficulties for affected children in school and work could be avoided.