

DNA sequencing is a molecular genetic method that results in data about sequence and type of nucleotides present in a given sample of deoxyribonucleic acid (DNA), a molecular carrier of genetic information. These data are frequently of a crucial value for many fields; research, medicine, industry, criminalistics or others. During a long period of time almost all the sequencing was performed using a method invented by Frederick Sanger in the 70's, a technique that uses modified nucleotides that once incorporated into a DNA strand prevent this from further elongation. DNA synthesis in presence of such nucleotides leads to a formation of a mixture of fragments of different length that are electrophoretically separated by length and the sequence is read from the resulting gel. Since the principle of this method entails some inherent drawbacks (e.g. low throughput and coverage) a significant effort is made lately to develop alternative sequencing approaches. These methods collectively referred to as next-generation sequencing (NGS) use several technologies in order to overcome the limitations of the Sanger sequencing.

This thesis discusses the most important NGS methods and focuses on their possible application for sequencing of immunoglobulin and T-cell receptor gene rearrangements, an area of undisputable clinical relevance for fields like immunology or hematology.