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## The use of new technology to improve genetic testing

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**Abstract:** Molecular confirmation of a clinical diagnosis of an inherited disease or of congenital malformations is of paramount importance for patients and their families. It is the conclusion of the differential diagnostic process, and provides information on the prognosis, in some cases on the therapeutic options, and on the recurrence risk. Currently, targeted sequencing of gene (s) of interest is the preferred approach for searching for small pathogenic mutations. The work presented in this thesis describes the application of new techniques for detecting small variations (mutations) in genomic DNA that underlie various disorders. These techniques include High Resolution Melting Curve Analysis (HR-MCA) followed by Sanger sequencing, targeted, X-exome and whole exome capture followed by Next Generation Sequencing (NGS). We have optimized, tested and applied the different new molecular techniques mentioned above to 1) faci litate the detection of disease causing mutations in several disorders with suspected Mendelian inheritance, 2) to speed up the identification of disease genes, 3) to provide a systematic tool for classifying previously intractable genetic diseases