MOLECULAR DIAGNOSTICS – MONOGENIC AND MULTIFACTORIAL DISEASES

Jadranka Sertic[a]
[a]University of Zagreb, Croatia

A program of molecular diagnostics has developed into a principal part of laboratory medicine over the past 20 years. In recent years, there is a phenomenon of rapidly growing number of diagnostic laboratories and an increasing number of genetic tests. Molecular testing laboratories use a wide variety of technologies and equipment and, according to best practice recommendations participate in external quality assessment. Molecular diagnostics of genetic diseases are advances in medicine for precision diagnosis, targeted individualized therapy and health care leading from intervention to prevention. Monogenic diseases have a high degree of inheritance, while in multifactorial diseases such as metabolic disorders and cardio- cerebrovascular diseases there is genetic heterogeneity. For a better understanding of the molecular basis of the disease, it is necessary to investigate the genetic mutations, because pathogenic mutations identify disease subtypes, which is crucial for accurate diagnosis and pharmacotherapy that modifies the course of the disease or refers to a surgical procedure. Translational research leads to molecular genetic diagnosis of monogenic diabetes, cystic fibrosis, haemochromatosis, alpha-1 antitripsin deficiency, multiple endocrine neoplasia, spinal muscular atrophy, myotonic dystrophy, Freidriech's ataxia, obesity, metabolic syndrome and stroke. The European Molecular Quality Network, Instand and Reference Institute for Bioanalytics harmonize molecular diagnostics, including the accuracy of genotyping and interpretation of the results.

Biography

Jadranka Sertic graduated from the Faculty of Chemical Engineering and Biotechnology, University of Zagreb, completed her Master's degree and PhD at the Zagreb University School of Medicine. She then specialized in Medical Biochemistry and Molecular Genetics at the Max-Planck-Institute of Biology, Tübingen. She is interested in laboratory medicine and molecular-genetic and biochemical approaches to hereditary diseases. Presently, she has been working at the Clinical Hospital Center Zagreb and Zagreb University School of Medicine.

jadranka.sertic@kbc-zagreb.hr

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