The Usage of Genetic Technologies in Endocrine Diseases

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In recent years, the genetic molecular methods are quickly replacing the classical method of measuring the hormone levels in clinical diagnosis and the studies related to endocrine diseases.

With the dazzling improvements in cell biology and molecular genetic technologies, the functions of hormone synthesis, secretions and regulations at cell levels become clearer. Today, the new technologies and genetic tests are being used routinely for diagnosis, follow-up and treatment of endocrine diseases along with scientific studies. It is mandatory to know and use these technologies, which provide convenience in daily practice of endocrinology doctors like for other branches of medicine and for the benefit of the patients. Usage of these powerful technologies in the field of endocrinology made it possible to investigate the molecular patterns and pathophysiology, to exhibit the genetic reasons behind, to describe the carriers, to give genetic consultancy and to do prenatal screening of the frequently seen endocrinological diseases which results in finding of new modalities of treatment to make it possible to cure these diseases. The improvements in computer and internet technologies made it easier to sequence the genome, which allowed the storage of the genetic data and its effortless use by the scientific people throughout the world. In 1980's, Genetic deformities were identified with karyotyping at the chromosomal level only. With this method, the number anomalies of chromosomes or big anomalies, namely bigger changes in the genome have only been acknowledged. Later on, in 1990's, methods such as fluorescence in situ hybridization and techniques which can identify the microdeletion/microduplications were developed and the diagnosis of diseases like Angelman syndrome and Prader-Willi syndrome became possible. In the last 10 years, there have been important developments in DNA and RNA sequencing technology. In the past, the genes of diseases that were thought to be the pre-diagnosis would have only been sequenced today, with the help of new generation sequence analysis technologies, the sequencing of lots of genes altogether (targeted sequence analysis) or the sequencing of the exons of the coded genes or sequencing of whole genome became possible. With this technology, RNA can also be sequenced and functional studies can be done as a result. The usage of these new methods is also becoming cheaper. In many monogenic endocrine diseases, the responsible gene is known and the identification of these genes is possible. Molecular tests can be basic diagnostic tools, however sometimes, they can be used to confirm the diagnosis or for screening. For example, in multiple endocrine neoplasia type 2 (MEN2), the mutations in the related genes are known before and as a result of this, prophylactic thyroidectomy is performed and thus molecular diagnosis is more important than any other diagnostic tests. Even though no precautions like this are taken before in *MEN1*, determination of the absence of a mutation in a patient will prevent to do unnecessary screening tests for several malignancies. Although the biochemical tests are prior to genetic tests in patient management and clinical diagnosis in 21-hydroxylase defect, the most common reason of congenital adrenal hyperplasia, when the prenatal diagnosis and treatment are in question, molecular testing becomes important. For diagnosis of MODY, molecular genetic tests play an important role in treatment planning and determining the prognosis. For polygenetic multifactorial endocrine diseases like type 1 and type 2 diabetes, the studies using the single nucleotide changes in genome by looking different loci shows that the risk ratio can be calculated. In endocrinological diseases, there are various molecular genetic tests performed by using either DNA or RNA. The tests should be chosen according to disease or to situation of the patient. With new generation sequence analysis, it is possible to investigate and diagnose a lot of genetic diseases with only one test by using the sequence of whole genome. As our knowledge about genome increases, application of these new technologies will become widespread and get easier.

Key words: Endocrine disorders, genetic technologies