Molecular Diagnosis of Familial Hypercholesterolemia

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Abstract

Background and objectives: Familial hypercholesterolemia (FH) is an autosomal disorder characterized by increased levels of total cholesterol and low density lipoprotein cholesterol. The FH clinical phenotype has been associated with increased risk of coronary heart disease and premature death. The mutation in LDLR gene in most cases is responsible for FH phenotype. Furthermore, other gene mutations such as apolipoprotein B- gene may cause similar results. Preliminary research indicates that the FH phenotype is also influenced by other genetic and environmental Factors; therefore, routine clinical analysis such as total cholesterol and LDL-C levels in serum, for early diagnosis and treatment, are not sufficient. Molecular diagnostic investigations, because of high specifity and sensitivity of near %100, administered for determining the prevalent mutations in LDLR (and probably other genes) are needed for exact diagnosis and accurate therapy. Currently, PCR-SSCP and southern blotting techniques are among the common techniques that could detect major mutations in gene.

Because of wide diversity in kinds of mutations in LDLR gene, we recommend, first, determining the proband's mutation and kinds of mutation, then, performing routine test based on type of mutation.

Key words: Familial hyperlipoproteinemia, LDL-R gene molecular diagnosis, mutation, Molecular Diagnostic Method