Research in Pharmaceutical Sciences, 2012;7(5)
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Molecular diagnosis of familial hypercholesterolemia

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Background and Aims: 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 mutations in LDLR gene in most cases is responsible for FH phenotype, however other gene mutations such as apolipoprotein B gene may cause similar abnormalities. Preliminary research indicates that the FH phenotype is also influenced by other genetic and environmental Factors so.

Routine clinical analysis such as total cholesterol and LDL-C levels in serum for early diagnosis are not sufficient and molecular diagnosis for prevalent mutations in LDLR (and probably other genes) is needed for determining exact criteria for disease, will approve the physician's therapy strategy. (specifity and sensitivity near %100). For current the used methods are PCR-SSCP and southern blotting techniques that could detect major mutations in gene.

Because of wide range diversity in kind of mutations in LDLR gene, it is reasonable that at first to determine the proband's mutation and after clearing the current site of mutation, performing site specific molecular population specific analysis by routine test based on type of mutation.

Keywords: Familial hyperlipoproteinemia; LDL-R gene molecular diagnosis; Mutation; Molecular diagnostic method