Diagnostic Assays For a Polymorphism in the SCARB1 Gene That Affects Expression and Function of the Receptor

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Technology description

Unmet Need:
High density lipoproteins (HDL) are considered good or healthy cholesterol that can protect against heart disease. However, there are people who have health problems despite having desirable levels of HDL. There are a number of factors that can contribute to the blood levels of HDL, including genetic problems and lifestyle choices. There is a need to develop methods and accompanying reagents that can be used to better assess an individual's susceptibility of developing cardiovascular disease.

Technical Overview:
JHU researchers have discovered that a sizeable number of people (approximately 34%) with desirable levels of HDL (defined by the National Cholesterol Education Program guidelines as being above 60 mg/dl) have a variation within a particular gene that leads to lower levels of a protective protein in their cells. They have identified. Specifically, they have identified a single nucleotide polymorphisms (SNP) as well as methods and kits useful in determining whether a subject is at increased risk for developing a cardiovascular disease by screening for the presence of a SNP in the scavenger receptor class B type I (SR-BI) gene of a subject.

The SNPs place the subject at greater risk of developing elevated levels of high density lipoprotein and cardiovascular disease. This invention may be developed into a diagnostic assay that identifies individuals who have a deficiency of an important protective protein that can lead to problems with heart disease and infertility.

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