New Method for Quantification of Allele-Specific RNA Expression, that Can be Used for Detection of Various Genetic Disorders

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Technology description

Summary

Available for licensing and commercial development is a new method for quantification of allele-specific RNA expression. This invention describes methods for simultaneously detecting the levels of expression of a plurality of different RNA transcripts expressed from a gene of interest in a subject or a cell. This is a simple assay to validate and quantify allele-specific silencing, by applying a combination of a fluorescent primer/probe set that specifically recognizes the targeted allele where the probe is labeled with one fluorophore, and a primer/probe set that specifically recognizes the normal allele, where the probe is labeled with another fluorophore in the same reaction tube. Furthermore, this method can be run on most real time PCR machines and requires very small amounts of RNA, less than 100 ng. This novel method, by comparing alleles within the same gene, expands on current real time PCR methods which compare one gene with another gene.

The invention also describes methods for validating the effectiveness and specificity of allele-specific siRNAs, kits for performing such assays, as well as methods for diagnosis of autosomal-dominant disorders, in which mutations in one allele result in a disease phenotype, such as Hutchinson-Gilford progeria, incontinentia pigmenti, neurofibromatosis, myotonic dystrophy, sialuria, Machado-Joseph disease, spinocerebellar ataxia, frontotemporal dementia, amyotrophic lateral sclerosis, slow channel congenital myasthenic syndrome, spinobulbar muscular dystrophy, as well as compound heterozygous autosomal recessive disorders. Other diseases that can be diagnosed include diabetes, cystic fibrosis, homocystenuria, Hermansky-Pudlak syndrome, cystinosis, Zellweger syndrome, beta-thalassemia, alkaptonuria, and cancer.

A variety of diseases appear to be mediated or accompanied by aberrant expression of one allele, often a mutant of a gene. Such differences in allelic expression can serve as the basis for diagnostic test for such conditions, and the ability to specifically silence the expression of detrimental alleles could be a therapeutic method for treating the disease, hence this novel method has very wide applications.

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