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Genotyping Assays of Single Nucleotide Polymorphism

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Description

Single nucleotide polymorphisms are the most common type of genetic variation found in humans. Each SNP represents a variation in a single DNA building block known as a nucleotide. In a specific stretch of DNA, for example, an SNP may replace the nucleotide cytosine (C) with the nucleotide thymine (T). SNPs are found naturally throughout a person's DNA. On average, they occurs nearly once every 1,000 nucleotides, implying that a person's genome contains 4 to 5 million SNPs. These variations occur in a large number of people; to be classified as an SNP, a variant must be found in at least 1% of the population. SNPs are distinct from substitution variants, which involve the replacement of one DNA building block (nucleotide) with another. Substitution variants usually cause disease and are found in less than 1% of any population. Furthermore, SNPs are distinct from copy number variants (CNVs), which occur when an entire gene (or other large section of DNA) is duplicated or deleted. SNPs are most commonly found in the DNA between genes [1-3].

They can serve as biological markers, assisting scientists in identifying genes linked to disease. SNPs that occur within a gene or in a regulatory region near a gene may play a more direct role in disease by affecting gene function. The majority of SNPs have no impact on health or development. However, some of these genetic differences have proven to be extremely important in the study of human health. SNPs can predict a person's response to certain drugs, susceptibility to environmental factors like toxins, and risk of developing diseases. SNPs can also be used to track disease-associated genetic variants' inheritance within families. SNPs associated with complex diseases such as heart disease, diabetes, and cancer are being studied. SNP arrays are high-density oligo arrays containing probes for SNPs located throughout the entire genome. These arrays are scanned, and SNP genotypes are called based on the fluorescence and the ratio of hybridization intensities for the two SNP alleles. The best known SNP array is a commercial array known as karyomapping. This is commercialized as a comprehensive, robust, off-the-shelf method for linkage-based testing of almost any single-gene disorder [4,5].

The analysis of several hundred thousand SNPs throughout the genome in the parents yields a set of informative SNP markers for each of the four parental chromosomes. The phase of the alleles for each informative SNP locus along each chromosome, as well as the linkage of the risk alleles with the parental chromosomes, can then be determined using the genotype of a relative with known disease status. The main advantage of this platform is that it can be applied to any familial single-gene disorder or any combination of loci within the chromosome regions covered by informative SNP loci, eliminating the need to develop patient- or disease-specific tests. Currently, PGT-M can determine both monogenic diagnosis and aneuploidy detection (along with HLA haplotyping) from the same sample. As a result, a single assay that uses the same platform to detect both chromosomal and monogenic disorders is preferable.

Because karyomapping defines distinct sets of SNP markers for each of the four parental chromosomes, it is such a method because it allows for accurate identification of the region of interest containing the mutation as well as simultaneous high-resolution molecular cytogenetic analysis. Meiotic trisomies are distinguished by the presence of both haplotypes from one parent in chromosomal segments resulting from the inheritance of two chromosomes with different recombination patterns, in addition to a single haplotype from the other parent. Furthermore, the absence of one of the parental haplotypes can be used to identify monosomies or deletions.

Conflict of Interest

None.

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