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An Editorial on Single Nucleotide Polymorphism Genotyping Tests

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Description

The most prevalent sort of genetic variation in humans is single nucleotide polymorphism. Each SNP is a change in a single nucleotide, the basic unit of DNA. For instance, an SNP may change the nucleotide cytosine (C) to the nucleotide thymine in a particular length of DNA (T). SNPs occur naturally all over a person's DNA. They occur roughly once every 1,000 nucleotides on average, which suggests that a person's genome has 4 to 5 million SNPs. Many people have these variants; nevertheless, for a variant to be considered an SNP, it must be present in at least 1% of the population. SNPs differ from substitution variations, which occur when one DNA building block (nucleotide) is swapped out for another. Less than 1% of any group has substitution variations, which typically cause disease. SNPs differ from copy number variants (CNVs), which take place when a whole gene (or other significant piece of DNA) is duplicated or deleted. SNPs are typically located between genes in the DNA [1-3].

They can act as biological markers, helping researchers find genes associated with disease. SNPs that influence gene function and are found within or close to regulatory regions of genes may have a more direct impact on disease. The vast majority of SNPs have no effect on growth or health. But some of these genetic variations have turned out to be crucial for understanding human health. SNPs can forecast a person's response to specific medications, vulnerability to chemicals in the environment, and likelihood of contracting diseases. SNPs can also be used to monitor the inheritance of disease-related genetic variations within families. SNPs linked to complicated illnesses like cancer, diabetes, and heart disease are being researched. High-density oligo arrays called SNP arrays contain probes for SNPs spread out over the entire genome. Based on the scanning of these arrays and the ratio of hybridization intensities for the two SNP alleles, SNP genotypes are identified. The commercial SNP array known as karyomapping is the most well-known. This is marketed as a complete, reliable, and readily available technique for linkage-based testing of virtually any single-gene condition [4,5].

For each of the four parental chromosomes, a set of useful SNP markers was produced after the analysis of several hundred thousand SNPs found across the parents' genomes. The genotype of a relative with a known illness status can then be used to establish the phase of the alleles for each relevant SNP location throughout each chromosome as well as the linkage of the risk alleles with the parental chromosomes. This platform's key benefit is its ability to use any combination of loci inside the chromosome areas covered by informative SNP loci or any familial single-gene problem, negating the need to create patient- or disease-specific tests. Currently, PGT-M may identify monogenic diagnosis and aneuploidy detection from the same sample (together with HLA haplotyping). Therefore, it is better to utilise a single assay that uses the same platform to identify both chromosomal and monogenic diseases.

In order to accurately identify the region of interest containing the mutation and perform high-resolution molecular cytogenetic analysis concurrently, karyomapping defines different sets of SNP markers for each of the four paternal chromosomes. Meiotic trisomies are recognised by the presence of a single haplotype from the other parent in addition to both haplotypes from one parent in chromosomal portions arising from the inheritance of two chromosomes with different recombination patterns. Furthermore, monosomies or deletions can be detected when one of the parental haplotypes is absent.

Conflict of Interest

The author declares that there is no conflict of interest associated with this paper.

References

1. Livak, Kenneth J., S. J. Flood and Jeffrey Marmaro, et al. "Oligonucleotides with fluorescent dyes at opposite ends provide a quenched probe system useful for detecting PCR product and nucleic acid hybridization." *Genome Res* 4 (1995): 357-362.
2. Alderborn, Anders, Anna Kristofferson and Ulf Hammerling. "Determination of single-nucleotide polymorphisms by real-time pyrophosphate DNA sequencing." *Genome Res* 10 (2000): 1249-1258.
3. Nyrén, Pettersson, Bertil Pettersson and Mathias Uhlén. "Solid phase DNA minisequencing by an enzymatic luminometric inorganic pyrophosphate detection assay." *Anal Biochem* 208 (1993): 171-175.
4. Pastinen, T., J. Partanen and Ann-Christine Syvänen. "Multiplex, fluorescent, solid-phase minisequencing for efficient screening of DNA sequence variation." *Clin Chem* 42 (1996): 1391-1397.
5. Tong, Anthony K, Zengmin Li and Gregg S. Jones, et al. "Combinatorial fluorescence energy transfer tags for multiplex biological assays." *Nat Biotechnol* 19 (2001): 756-759.

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