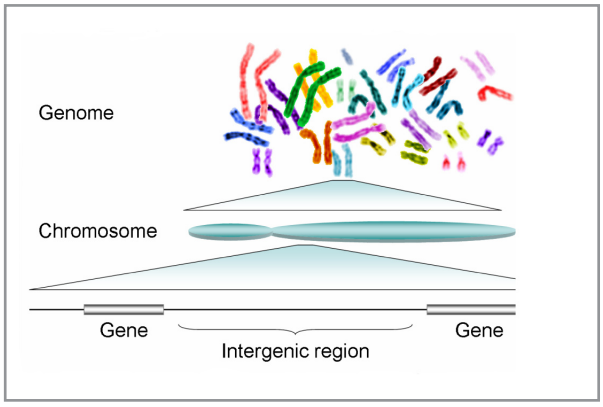
BCCGN
British Columbia Clinical Genomics Network

SINGLE NUCLEOTIDE POLYMORPHISM (SNP) GENOTYPING

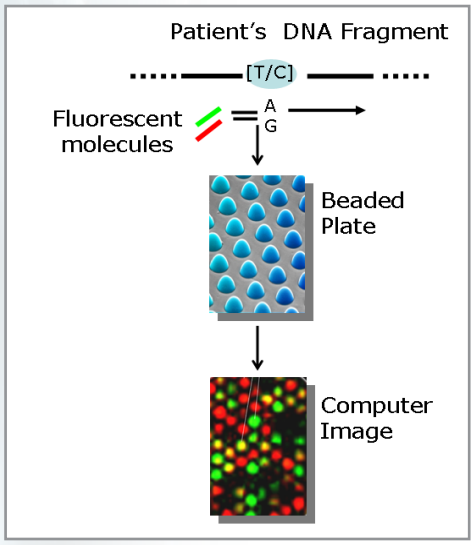
GENES 101

The 23 pairs of chromosomes in our genome contain approximately 30,000 genes. A gene is made up of a long molecule of double stranded DNA, which in turn is a nucleotide sequence consisting of four bases (A, T, C, or G). Genes encode all the biological information that parents pass on to their children. Sometimes a mistake occurs that results in a deletion, duplication or rearrangement of some of this genetic material such as a whole chromosome or part of a chromosome, causing a genetic disorder. Smaller alterations within genes, or the deletion or substitution of a single nucleotide also occur. If this change is at a critical position, it can affect the way a gene is expressed and thus can cause disease.



SINGLE NUCLEOTIDE POLYMORPHISM (SNPs)

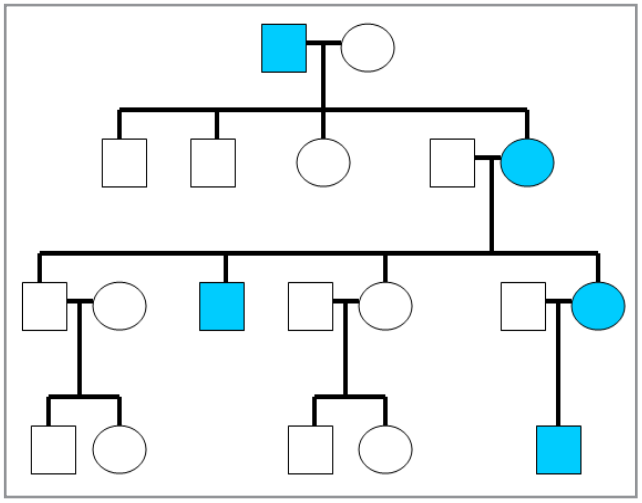
Our DNA sequences are 99.9% identical to that of another person. Of the remaining 0.1%, the most common variation at a single base is called a single nucleotide polymorphism or a SNP. SNPs occur about every 1000 bases so each of us has about 5 million differences like this in our genome. These changes are normal and do not cause disease. These SNPs can be used as markers in SNP genotyping tests.



SNP GENOTYPING

A SNP genotyping test looks for common variations that may be "linked" (located very close) to a gene that has been altered. To do this, an individual's DNA is cut up into small fragments. A fluorescent molecule recognizes a specific SNP on each fragment and binds to it. The fluorescent DNA fragments are added to a 96-well plate in which each well contains 50,000 beads. Each bead is already coated with a single specific piece of DNA whose location is known on the genome. The individual's labeled DNA fragments will find their complementary match on the beads, resulting in the generation of a fluorescent signal. Computer analysis of the fluorescent signals determines an individual's SNP genotype at each location, thus generating a SNP genotype for the entire genome.

WHAT CAN I USE THIS TECHNOLOGY FOR?



It can be applied to a variety of different studies. It can be used to track a SNP that is "linked" to a gene of interest. If there are several affected family members in one family, then the same SNP will follow the disease gene as it gets passed from one family member to another. Knowing which SNP is "linked" to the gene, and where that SNP is located in the genome can help determine the location of the disease gene. SNP genotyping can also be used to look for SNPs that "associate" with particular disease genotypes such as heart disease, Alzheimer's Disease and diabetes using large numbers of unrelated individuals. Clinicians involved in studies that identify SNPs associated with a particular disease or risk can help achieve a greater understanding of the condition and better therapies for their patients.

HOW CAN I GET INVOLVED?

If you are a clinician interested in genomic research, please contact us for more information or visit our web site to submit a research proposal

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