Genome generation

Glossary of terms

Chromosome
An organism's DNA is packaged into chromosomes. Humans have 23 pairs of chromosomes including one pair of sex chromosomes. Women have two X chromosomes and men have one X and one Y chromosome.

Dominant (see also recessive)
Genes come in pairs. A dominant form of a gene is the "stronger" version that will be expressed. Therefore if someone has one dominant and one recessive form of a gene, only the characteristics of the dominant form will appear.

DNA
DNA is the long molecule that contains the genetic instructions for nearly all living things. Two strands of DNA are twisted together into a double helix. The DNA code is made up of four chemical letters (A, C, G and T) which are commonly referred to as bases or nucleotides.

Gene
A gene is a section of DNA that is the code for a specific biological component, usually a protein. Each gene may have several alternative forms. Each of us has two copies of most of our genes, one copy inherited from each parent. Most of our traits are the result of the combined effects of a number of different genes. Very few traits are the result of just one gene.

Genetic sequence
The precise order of letters (bases) in a section of DNA.

Genome
A genome is the complete DNA instructions for an organism. The human genome contains 3 billion DNA letters and approximately 23,000 genes.

Genomics
Genomics is the study of genomes. This includes not only the DNA sequence itself, but also an understanding of the function and regulation of genes both individually and in combination.

Genotype
A genotype describes the particular versions of genes in an individual. For example someone could have a genotype that means they are intolerant of lactose or have an increased risk of developing a particular disease.

Genotyping test
Genotyping tests analyse single letter changes (SNPs) in an individual's DNA to discover which variants of particular genes that person has. Some commercially available genotyping tests offer an extensive analysis which gives background on ancestry, tells you whether you are a carrier for a range of genetic diseases, indicates certain disease risks and shows you the genetics behind traits such as eye colour and perception of bitter tastes.

Mutation
A mutation is a change to the DNA sequence. Mutations can take a number of forms including single letter changes (SNPs), deletion or duplication of sections of DNA and joining together of different sections of DNA.

Most mutations have no effect at all but some can be beneficial or harmful.

Pharmacogenomics
Pharmacogenomics is an emerging area of medicine. It involves analysing entire genomes, across groups of individuals, to identify the genetic factors influencing responses to a drug. Pharmacogenomics aims to produce more effective, personalised medicines.

Predisposition
If someone has a genetic predisposition to a disease it means that they have a specific variant of a gene or genes that increases their risk of developing a particular disease (compared to the average population). Although people with a predisposition to a genetic disease are at an increased risk, there is no guarantee that they will develop the disease. Similarly, those who do not have the predisposition have no guarantee that they will not develop the disease.

Recessive (see also dominant)
Genes come in pairs. A recessive gene is the "weaker" of a pair. The recessive characteristics will only be seen if an individual has a pair made of two recessive genes. If a person has one recessive gene and one dominant gene, the recessive characteristic will be overwhelmed and only the dominant characteristic will be seen.

Risk
Disease risks are often described as a percentage. If someone has an 80% risk of a disease this means that 80 out of 100 people in their situation develop the disease.

SNPs
SNPs (pronounced "snips") are single letter changes in a DNA sequence. The human genome contains an estimated 15 million SNPs. Some SNPs are linked to differences in particular traits but most do not cause observable differences between people. SNP is short for single nucleotide polymorphism.

For further information, see the Your Genome website's glossary of terms at:
http://www.yourgenome.org/glossary/