



# Would you want your genome sequenced?



As an experiment, some of our SciBar participants prepared this glossary, independently of Andrew Read. You may find it informative to consider these concepts and visit the website. Your comments on this experiment are most welcome.

**Allele** every gene comes in different versions called alleles, which produce different characteristics. So a gene for eye colour may come in a 'blue type' allele and a 'brown type' allele. Since each body cell has two copies of every chromosome, one from the mother and one from the father, there are two copies of each gene, which may be the same or different alleles.

**Base** building blocks of DNA: adenine (A), cytosine (C), guanine (G) and thymine (T)

**Carrier** someone who has inherited a mutation for a genetic disorder but do not themselves have the disorder. They have only one copy of the recessive allele that causes the disorder.

**Chromosomes** structures found in the nucleus of the cell that carry the genetic information

**DNA** (deoxyribose nucleic acid) the molecule that encodes genetic information

**DNA sequence** the order or sequence of all the bases in a gene or genome, using the letters A, C, G, T

**Dominant allele** version of a gene (allele) that has an effect even when only one such allele from either parent is present

**Environmental factors** factors in the environment that may have an effect on the development of disease, such as diet

**Gene** found on chromosomes; a section of DNA that determines an inherited characteristic, such as eye colour or susceptibility to a certain disease

**Genetic disorder** a disorder that can be passed on from parents to children in faulty genes. The alleles for this gene can be dominant or recessive. However few disorders have a single-gene cause: many result from a combination of genes and their interaction with lifestyle and environmental factors.

**Genetic testing** until recently, referred to testing for single-gene defects, e.g. for adult-onset diseases such as Huntington's disease. There are now susceptibility genes identified for Alzheimer's disease and many cancers, leading to the possibility of telling people if they are at risk for these diseases.

**Genome** all of the genetic information (DNA) of an organism, as a list in order of every base. Potentially has information about your risk of developing certain diseases in future.

**Genomic medicine** the use of genetic information to determine risk of developing a disease, diagnosis and the best drugs for treating particular patients

**Human genome project** a project to work out the order of all of the base pairs of the human genome. Although each human being has a unique DNA sequence the project shows that everyone has at least 99.9% of their DNA in common.

**Marker** a gene or other portion of DNA whose inheritance can be followed

**Mutation** change in the DNA sequence

**Recessive allele** version of a gene (allele) that will only have an effect if a recessive allele is inherited from both parents

## Useful weblinks:

Making Sense of Testing, [www.senseaboutscience.org.uk/pdf/makingsenseoftesting.pdf](http://www.senseaboutscience.org.uk/pdf/makingsenseoftesting.pdf)