

Glossary of terms for public health genomics

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In this Glossary the secretariat of PHGEN has compiled some of the terms relevant to Public Health Genomics. A more extensive version of this glossary can be found on the PHGEN-website www.phgen.eu.

A

Assessment

The systematic collection, assembly, analysis and dissemination of information (including human genome epidemiology information) on the health of the community.

Assurance

Assurance constitutes that genetic information is used appropriately and that genetic tests and services meet agreed-upon goals for effectiveness, accessibility and quality.

B

Biobank

Biobanks are databases where biological material such as DNA, blood- or tissue samples together with background information about the donors is stored.

C

Carrier screening

Testing routinely offered to unaffected individuals for genetic conditions in order to detect those at increased risk of having affected children. Usually refers to carriers of recessive conditions, e.g. beta thalassaemia or cystic fibrosis.

Complex trait

Trait that has a genetic component that does not follow strict Mendelian inheritance. May involve the interaction of two or more genes or gene-environment interactions.

D

Determinant of health

Any factor, whether event, characteristic, or definable entity, that brings about change in a health condition or other defined characteristic (e.g. Physical and social environmental factors, biological and genetic factors, factors relating to the health care system, lifestyle factors).

DNA (Desoxyribonucleic acid)

The molecule that encodes genetic information. DNA is a double stranded molecule held together by weak bonds between base pairs of nucleotides. There are four types of nucleotides in DNA (the four building blocks). Each nucleotide consists of a base, a sugar (desoxyribose) and a phosphate group. The four nucleotides in DNA consist of the bases adenine (A), guanine (G), cytosine (C), and thymine (T).

E-F

Empowerment

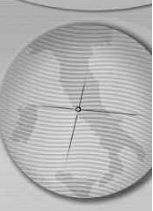
In health promotion, empowerment is a process through which people gain greater control over decisions and actions affecting their health.

Epidemiology

The study of the distribution and determinants of health-related states or events in specified populations, and the application of this study to the control of health problems.

Epigenetic

A factor or mechanism that changes the expression of a gene or genes without changing their DNA



sequence. In more general terms, an epigenetic factor is something that changes the phenotype without changing the genotype.

G

Gene

A part of the DNA molecule of a chromosome which encodes (directs the synthesis of) a protein

Genetic counselling

Provides patients and their families with accurate information about a particular genetic test and the implications of the results.

Genetic epidemiology

A field of research in which correlations are sought between phenotypic trends and genetic variations across population groups.

Genetic heterogeneity

Refers to diseases, conditions or other characteristics that appear similar but whose genetic basis is different in different populations or individuals.

Genetic literacy

Enabling professionals and the population to understand the role of genetics in health and to make the best use of genetic information for preventing disease, improving health and increasing the quality of life for individuals with special health needs.

Genetic test

A test to detect the presence or absence of, or change in, a particular gene or chromosome. Technically a Genetic Test may be any test, that yields genetic data, unambiguously underlying DNA information, either germline or somatic, and where the nature of the test is not important, but the genetic information derived.

Genome

The total sum of the genetic material present in a particular organism.

Genomics

The study of genes and their function

H

Health Technology Assessment (HTA)

Technology assessment in health care is a multidisciplinary field of policy analysis. It studies systematically the medical, social, ethical, and economic implications of development, diffusion, and use of health technology.

Health Need Assessment (HNA)

HNA is a systematic method for reviewing the health issues facing a population, leading to agreed priorities and resource allocation that will improve health and reduce inequalities.

Heritability

The degree to which a characteristic is determined by genetics

I-L

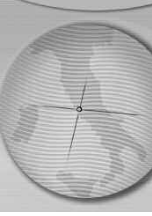
Informed consent

Informed Consent refers to a process by which a subject voluntarily confirms his or her willingness to participate in a particular trial after having been informed of all aspects relevant to the subject's decision.

M

Monogenic Disease

A disease whose pathology results from the presence of a particular allele of a gene, either in a heterozygous or homozygous state.

**Multifactoriell disease**

A disease whose pathology is dependant on the complex interplay of several genetic and environmental factors.

Mutation

A change in a gene or chromosome that causes a disorder or the inherited susceptibility to a disorder.

N-O**Nutrigenomics**

Nutrigenomics is the science that examines the response of individuals to food compounds using post-genomic and related technologies (e.g. genomics, transcriptomics, proteomics, meta-bolnomics etc.).

P-Q**Penetrance**

The likelihood that a person carrying a mutation will develop the characteristics caused by that mutation.

Phenotype

The observable traits of an organism. The phenotype results from the combination of genetic and environmental factors.

Policy Development

The formulation of standards and guidelines in collaboration with stakeholders, which promote the appropriate use of genetic information and the effectiveness, accessibility and quality of genetic tests and services.

Polygenic disorder

A genetic disorder resulting from the combined action of alleles of more than one gene (e.g., heart disease, diabetes, and some cancers). Although such disorders are inherited, they depend on the simultaneous presence of several alleles; thus the hereditary patterns are usually more complex than those of single-gene disorders.

Polymorphism

Variation in a region of DNA sequence among different individuals; the variation should be present in at least 1-2% of the population to be considered a polymorphism.

Predictive testing

Genetic testing carried out to detect a particular gene mutation before the onset of symptoms, offered to those at risk of a disorder, e.g Huntington disease.

Predictive Values

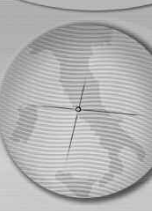
In screening and diagnostic tests, the probability that a person with a positive test is a true positive (i.e., does have the disease) is referred to as the "predictive value of a positive test. The predictive value of a negative test is the probability that a person with a negative test does not have the disease. The predictive value of a screening test is determined by the sensitivity and specificity of the test, and by the prevalence of the condition for which the test is used.

Predisposition, genetic

The increased probability (compared to the general population) of developing a disease, due to the presence of one or more gene mutations. Genetic predispositions, unlike most 'single gene' genetic conditions, are modified by non-genetic influences, such as diet or smoking.

Prevention

In medicine, prevention is any activity by which an individual avoids the development of a disease (primary prevention), diagnosis a disease in an early stage or prevents its reoccurrence (secondary prevention), or avoids a disease's worsening and restores oneself to an optimal level of functioning (tertiary prevention).



Public Health

Public health is a social and political concept aimed at improving health, prolonging life and improving the quality of life among whole populations through health promotion, disease prevention and other forms of health intervention.

Public Health Genomics

Public Health Genomics can be described as the responsible and effective translation of genome-based knowledge and technologies for the benefit of population health.

Public Health Trias

Describes the three core functions of Public Health: Assessment, Policy Development and Assurance.

R

Risk factor

An aspect of personal behaviour or life-style, an environmental exposure, or an inborn or inherited characteristic, that, on the basis of epidemiological evidence, is known to be associated with health-related condition(s) considered important to prevent.

Risk communication

In genetics, a process in which a genetic counsellor or other medical professional interprets genetic test results and advises patients of the consequences for them and their offspring.

S-U

Screening, genetic

Carrying out a genetic test on a whole unselected population, or on all the members of a subset of the population (for example, people from a particular ethnic group, or pregnant women, or newborn infants).

Sensitivity

Sensitivity is the proportion of truly diseased persons in the screened population who are identified as diseased by the screening test. Sensitivity is a measure of the probability of correctly diagnosing a case, or the probability that any given case will be identified by the test.

Single-gene disorder

Hereditary disorder caused by a mutant allele of a single gene.

Specificity

Specificity is the proportion of truly nondiseased persons who are so identified by the screening test. It is a measure of the probability of correctly identifying a nondiseased person.

Stratification

The process by which groups are separated into mutually exclusive sub-groups of the population that share a characteristic: e.g. age group, sex, or socioeconomic status. It is possible to compare these different strata to try and see if the effects of a treatment differ between the sub-groups.

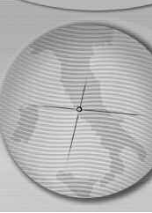
Susceptibility, genetic

Predisposition to a particular disease due to the presence of a specific allele or combination of alleles in an individual's genome.

V-Z

Validity, analytical

Refers to how well the test (in this case *genetic test*) measures the property or characteristic it is intended to measure. It can be defined as sensitivity, specificity and reproducibility of the tests measurement of a particular genetic variant.

**Validity, clinical**

Refers to the accuracy with which a test predicts the presence or absence of a certain disease or predisposition. Clinical validity data ideally should be based on strong epidemiologic study design and may be represented as the sensitivity, specificity or positive and negative predictive value of a test for a particular clinical outcome.

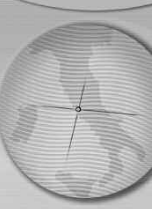
Resources*Websites:*

Genomicseducation. Glossary. <http://www.genomicseducation.ca/glossary/a.htm> (accessed september 2006).
 Glaxo Smith Kline. Genetics glossary. <http://genetics.gsk.com/glossary.htm> (accessed september 2006)
 Human Genome Project Information (HGPI). Genome glossary. http://www.ornl.gov/sci/techresources/Human_Genome/glossary/glossary.shtml (accessed september 2006).
 National Library for Health. Genepool Glossary of Clinical Genetics Terms. <http://www.library.nhs.uk/genepool/Page.aspx?pagename=GLOSSARY> (accessed september 2006).
 Public Health Genetics Unit. Glossary of genetic terms. <http://www.phgu.org.uk/pages/resources/glossary.htm> (accessed september 2006).
 The Cochrane Collaboration. Glossary of Cochrane Collaboration terms. <http://www.cochrane.org/resources/glossary.htm> (accessed september 2006)

Other Sources:

Beskow LM, Khoury MJ, Baker TG, Thrasher JF The Integration of Genomics into Public health Research, Policy and Practice in the United States. *Community Genetics* 2001;4:2-11.
 Genomics Working Group of the Science and Technology Task Force of the United Nations Millenium Project. *Genomics and Global Health*.
 IoM (Institute of Medicine). *The Future of Public Health*. Division of Health Care Services. Committee for the Study of the Future of Public Health. Washington D.C.: National Academy Press, 1988.
 Last JM. *A Dictionary of Epidemiology*. 4th ed. Oxford: Oxford University Press, 2001
 NHS Department of Health. *Our inheritance, our future*, 2003
 WHO. *Genomics and World Health - Report of the Advisory Committee on Health Research*. Geneva, 2002
 WHO. *Health Promotion Glossary*. Geneva, 1998

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