# BIOETHICS THESAURUS FOR GENETICS
# Alphabetical List of Keyword Descriptors and Genetics Tree

## 2010

**Appendix:**
**Examples of Genetics-Related Keyword Identifiers**

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## Introduction: Keyword Indexing of the GenETHX Database

With financial support from the National Human Genome Research Institute, the Bibliographers at the Bioethics Research Library at Georgetown University have updated and enriched the *Bioethics Thesaurus* with genetics terminology. In May 2007 they began indexing documents in the GenETHX database with these terms.

The 2010 *Bioethics Thesaurus for Genetics* contains 108 Keyword Descriptors (subject terms in a controlled indexing vocabulary) related to genetics. Ten of these terms are new to the 2010 Thesaurus. Some of the terms (e.g., CLINICAL GENETICS, GENETIC ANCESTRY, GENETIC DISCRIMINATION, GENETIC EPIDEMIOLOGY, GENETIC RELATEDNESS TIES, GENETIC RESOURCES, HUMAN GENOME DIVERSITY PROJECT, PERSONAL GENOMICS, and SYNTHETIC BIOLOGY) are, as far as we know, unique to any Thesaurus descriptor vocabulary.

All of the Keyword Descriptors in the Bioethics Thesaurus for Genetics have been annotated with the date they were introduced (DT), with explanatory Scope Notes (SN), and with Broader Term (BT), Narrower Term (NT), and Related Term (RT) cross-references to other Thesaurus terms, some in subject areas other than genetics. (Following Thesaurus convention, "+" after a Narrower or Related Term indicates that the term cited has more specific Narrower Terms under it.)

The Classification field (CL) indicates what number, or combination of numbers, from the Bioethics Research Library's Classification Scheme would have been assigned to the document. ("+" after a number indicates that a searcher might find any number starting with 15. that appears on the Classification Scheme.)

The alphabetical list of Keyword Descriptors in the 2010 *Bioethics Thesaurus for Genetics* is followed by an updated "Genetics Tree," which provides an easy-to-grasp three-page overview of the genetics terms in a hierarchical display. Searchers interested in a set of broader and narrower terms should "OR" the relevant terms together in a search strategy statement.

An appendix to this Thesaurus provides examples of genetics-related Keyword Identifiers (KWIs) that are not part of the controlled vocabulary of the *Bioethics Thesaurus*. They include the names of persons, corporate bodies, laws, court decisions, and geographic names.

So far, more than 3,850 documents in the GenETHX database have been indexed with Keyword Descriptors from the *Bioethics Thesaurus* and, as appropriate, with non-Thesaurus Keyword Identifiers, and Keyword Proposed Descriptors (terms under consideration for inclusion in future editions of the *Bioethics Thesaurus*). They have also been indexed with many non-genetics related Keywords from the full *Bioethics Thesaurus*.

The comprehensive *Bioethics Thesaurus* is available on the Web at http://bioethics.georgetown.edu/databases/bt/.

Search tips geared to searching the GenETHX database are available on the Web at http://bioethics.georgetown.edu/databases/GenETHX/genetips/index.html.

## KEYWORD DESCRIPTORS IN THE 2010 BIOETHICS THESAURUS FOR GENETICS

**ADULT STEM CELLS**
DT 2007
SN Undifferentiated cells found in a differentiated tissue that can renew themselves and differentiate (with certain limitations) to give rise to more specialized cells
BT Stem cells
RT Induced pluripotent stem cells
CL 18.7

**ALTERED NUCLEAR TRANSFER**
DT 2010
SN A proposed technological approach to obtain human pluripotent stem cells, which are the functional equivalent of human embryonic stem cells, without creating or destroying human embryos (ANT website, accessed 1/26/10). The process was originally proposed by William B. Hurlbut
BT Nuclear transfer techniques
RT Induced pluripotent stem cells
CL 18.7 and 15.1

**ANIMAL CLONING**
DT 2007 (was CLONING, 1974-2001)
SN Asexual single-parent reproduction of an animal, in which the offspring has DNA in the cell nucleus that is identical to the nuclear DNA of its parent
BT Cloning
CL 14.5 and 22.3

**APO-E GENES**
DT 2009
SN Genes that provide instructions for making a protein called apolipoprotein E which carries cholesterol and other fats through the blood to be processed. Some variations of the APO-E gene are associated with a genetic predisposition to disorders such as Alzheimer disease
UF apoE genes
apolipoprotein genes
BT Genes
CL 15.+

**BEHAVIORAL GENETICS**
DT 1974
SN The study of the effects of heredity on human behavior
BT Genetics
NT Genetic determinism
RT Psychiatric genetics
Sociobiology
XYY karyotype
CL 15.6

**BIOTECHNOLOGY**
DT 2008
SN The manipulation (as through genetic engineering) of living organisms or their components to produce useful, usually commercial, products (as pest resistant crops, new

bacterial strains, or novel pharmaceuticals; also, any of various applications of biological science used in such manipulation (Merriam-Webster Online Dictionary, accessed 1/17/08)
BT Genetic engineering
Technology
NT Synthetic biology
CL 15.+

**BRCA1 GENES**
DT 2008
SN Tumor suppressor genes, located on human chromosome 17, mutations of which are associated with a higher than average incidence of breast and ovarian cancer
BT Genes
CL 15.+

**BRCA2 GENES**
DT 2008
SN Tumor suppressor genes, located on human chromosome 13, mutations of which are associated with a higher than average incidence of breast and ovarian cancer
BT Genes
CL 15.+

**CANAVAN DISEASE**
DT 2010
SN An inherited disorder that causes progressive damage to nerve cells in the brain (Genetics Home Reference, accessed 1/20/10)
BT Central nervous system diseases
Genetic disorders
CL 15.+

**CHIMERAS**
DT 2007 (was HYBRIDS, 1974-2001)
SN The offspring of parents belonging to different species, varieties, or genotypes
UF Hybrids
BT Genetic engineering
CL 15.1 and 22.1

**CHROMOSOME ABNORMALITIES**
DT 1974
SN Defects in the structure of number of chromosomes resulting in structural aberrations or manifesting as disease (Online Medical Dictionary, accessed 11/25/08)
UF Aneuploidy
BT Congenital disorders
Genetic disorders
NT Fragile X syndrome
Klinefelter syndrome
Trisomy+
Turner syndrome
XYY karyotype
CL 15.+

**CLINICAL GENETICS**
DT 2007
SN Genetics applied to the diagnosis, prognosis, management, and prevention of genetic

diseases (Stedman's Online Medical Dictionary, 2007)
UF Medical genetics
BT Genetics
NT Gene therapy
Genetic counseling
Genetic services
Genetic testing
Preimplantation diagnosis
Prenatal diagnosis+
RT Genetic intervention+
CL 15.2 or 15.3 or 15.4

**CLONING**
DT 1974
SN Asexual single-parent reproduction, in which an offspring has DNA in the cell nucleus that is identical to the nuclear DNA of its parent
UF Human cloning
Embryo cloning (use CLONING and (EMBRYOS or EMBRYO RESEARCH+))
Reproductive cloning (use CLONING and REPRODUCTIVE TECHNOLOGIES)
Research cloning (use CLONING and EMBRYO RESEARCH+)
Therapeutic cloning (use CLONING and (STEM CELLS+ or STEM CELL TRANSPLANTATION))
BT Genetic techniques
Reproductive technologies
NT Animal cloning
RT Nuclear transfer techniques+
CL 14.5

**CYSTIC FIBROSIS**
DT 1981
SN An autosomal recessive genetic disease of the exocrine glands (MeSH, truncated)
BT Genetic disorders
CL 15.+

**DNA**
DT 2007 (was GENETIC MATERIALS, 1997-2001)
SN A nucleic acid that is the primary genetic material of all cells
UF Deoxyribonucleic acid
BT Genetic materials
RT DNA sequences
CL 15.1 or 15.8

**DNA FINGERPRINTING**
DT 1991
SN A genetic identification procedure in which band patterns of DNA (the DNA fingerprint) from one individual or an unknown individual are evaluated for similarities with those of a known individual
BT Forensic genetics
Genetic techniques
RT Genetic databases
Genetic testing
CL 15.1 and 1.3.5

**DNA SEQUENCES**
DT 1997
SN Base pairs arranged in linear order, whether in a stretch of DNA, a gene, a chromosome, or an entire genome
UF Base sequence
BT Genetic materials
RT DNA
Genes+
Genome+
CL 15.1 or 15.8

**DOWN SYNDROME**
DT 1974
UF Down's syndrome
Mongolism
Trisomy 21
SN A chromosome disorder associated either with an extra chromosome 21 or an effective trisomy for chromosome 21 (MeSH, truncated)
BT Trisomy
RT Mentally retarded persons
CL 9.5.3 (often and 15.2)

**DUCHENNE MUSCULAR DYSTROPHY**
DT 1974
UF Dystrophy, Duchenne muscular
Muscular dystrophy, Duchenne
SN One of nine types of muscular dystrophy, a group of genetic, degenerative diseases primarily affecting voluntary muscles (Muscular Dystrophy Association website, accessed 12/4/08)
BT Genetic disorders
CL 15.+

**EMBRYONIC STEM CELLS**
DT 2007
SN Primitive (undiffentiated) cells derived from a 5-day blastocyst that have the potential to become a wide variety of specialized cell types
BT Stem cells
CL 18.7

**EPIGENETICS**
DT 2008 kwds
SN Heritable changes in phenotype (appearance) or gene expression that are caused by mechanisms other than changes in the underlying DNA sequence (Wikipedia)
BT Genetic phenomena
CL 15.+

**EUGENICS**
DT 1974
SN The study of or belief in the possibility of improving the qualities of the human species or a human population, esp. by such means as discouraging reproduction by persons having genetic defects or presumed to have inheritable undesirable traits (negative eugenics) or encouraging reproduction by persons presumed to have inheritable desirable traits (positive eugenics) (based on the Random House Unabridged Dictionary, 2006)

UF Negative eugenics
Positive eugenics
BT Genetic intervention
RT Genetic enhancement
CL 15.5

**EVOLUTION**
DT 1974
UF Natural selection
SN The process of cumulative change over successive generations through which organisms acquire their distinguishing morphological and physiological characteristics (MeSH)
BT Genetic phenomena
RT Sociobiology
CL 15.1 and/or 3.2

**FORENSIC GENETICS**
DT 2008
SN The branch of genetics that deals with the application of genetic knowledge to legal problems and legal proceedings, often referring specifically to matching DNA from a suspect in a crime to samples taken from a crime scene
BT Forensic medicine
Genetics
NT DNA fingerprinting
RT Law enforcement
CL 15.1 and 1.3.5

**FRAGILE X SYNDROME**
DT 2008
SN A genetic disorder caused by a mutation of the FMR1 gene on the X chromosome. It is associated with a range of developmental problems, including cognitive or intellectual disabilities
BT Chromosome abnormalities
RT Mental retardation
CL 15.+

**GAUCHER DISEASE**
DT 2009; *was* GENETIC DISORDERS, 1974-2001
SN A rare genetic disorder that results in the accumulation of fatty molecules called cerebrosides
BT Genetic disorders
CL 15.+

**GENE POOL**
DT 1974
SN The complete assortment of genes present in the gametes of the members of a population that are eligible to reproduce (Encyclopedia of Genetics, 2004)
BT Genetic phenomena
RT Population genetics+
CL 15.+

**GENE THERAPY**
DT 1980
SN The treatment of genetic disorders through the introduction of properly functioning genes into the appropriate cells of an organism

UF Germline gene therapy (use GENE THERAPY and GERM CELLS)
Somatic gene therapy
BT Clinical genetics
Genetic engineering
RT Gene transfer techniques
Genetic enhancement
Genetic services
CL 15.4

**GENE TRANSFER TECHNIQUES**
DT 2007
SN Methods of introducing genes into an organism's cells, usually by a vector such as a modified virus
BT Genetic techniques
RT Gene therapy
Genetic enhancement
CL 15.+

**GENES**
DT 1997
SN Functional and physical units of heredity passed from parent to offspring. Genes are pieces of DNA, and most genes contain the information for making a specific protein (National Human Genome Research Institute Talking Glossary, 2007)
BT Genetic materials
NT APO-E genes
BRCA1 genes
BRCA2 genes
Transgenes
RT DNA sequences
CL 15.1 or 15.8

**GENETIC ANCESTRY**
DT 2007
SN Relationship to continental population groups through one's forebears
BT Population genetics
RT Pedigree
Pharmacogenetics
Racial groups+
CL 15.11

**GENETIC ASSOCIATION STUDIES**
DT 2010
SN The analysis of a sequence such as a region of a chromosome, a haplotype, a gene, or an allele for its involvement in controlling the phenotype of a specific trait, metabolic pathway, or disease (MeSH)
BT Genetic techniques
NT Genome-wide association studies
CL 15.1

**GENETIC CARRIERS**
DT 2007 (was CARRIERS, 1974-2001)
SN Individuals possessing a specified gene who are capable of transmitting it to offspring but who do not show its typical expression
UF Carriers

BT Genotype
RT Genetic disorders+
CL 15.2 or 15.3

**GENETIC COUNSELING**
DT 1974
SN The process whereby an expert in genetic disorders provides information about risk and clinical burden of a disorder or disorders to patients or relatives in families with genetic disorders as an aid to making informed and responsible decisions about marriage, children, early diagnosis, and prognosis (Stedman's Online Medical Dictionary, 2007)
BT Counseling
Clinical genetics
Genetic intervention
RT Directive counseling
Genetic information+
Genetic services
Genetic testing
Prenatal diagnosis+
CL 15.2

**GENETIC DATABASES**
DT 2007 (was DNA DATA BANKS, 1994-2001)
SN Collections of genetic information that are maintained for clinical, research, or law enforcement purposes
UF DNA data banks
DNA databases
BT Databases
Genetic information
RT Biological specimen banks+
DNA fingerprinting
Genetic privacy
Genetic research+
CL 15.1 and 1.3.12

**GENETIC DETERMINISM**
DT 1998
SN The theory that human character and behavior are determined solely or predominantly by the genes that comprise the individual's genotype, rather than shaped by genotype plus culture, social environment, and individual choice; also used for discussions of the degree to which genes determine physical phenotypes
UF Genetic reductionism
BT Behavioral genetics
Genetic phenomena
CL 15.6

**GENETIC DISCRIMINATION**
DT 2007 (was SOCIAL DISCRIMINATION, 1982-2001, or INSURANCE SELECTION BIAS, 1998-2001)
SN Unfair treatment of a person or group based on genetic information, which may be used, e.g., to deny or limit insurance or employment
BT Psychosocial genetics
Social discrimination
CL 15.+ and (8.4 or 9.3.1 or occasionally 16.3)

**GENETIC DISORDERS**
DT 1974
SN Pathological conditions caused by an absent or defective gene or by a chromosome aberration (American Heritage Dictionary of the English Language, 4th ed, 2000)
UF Genetic diseases, inborn
Hereditary diseases
Inborn genetic diseases
Inherited disorders
BT Genetics
NT Canavan disease
Chromosome abnormalities+
Cystic fibrosis
Duchenne muscular dystrophy
Hemoglobinopathies+
Hemophilia
Late-onset disorders+
Phenylketonuria
Tay Sachs disease
*Note: consult the MeSH vocabulary (under Genetic Diseases, Inborn) for additional genetic disorders*
RT Genetic carriers
Genetic predisposition
Growth disorders
Neural tube defects+
CL 15.+

**GENETIC DIVERSITY**
DT 1998
SN Genotypic differences among individuals and among population groups
UF Genetic variation
Variation, genetic
BT Genetic phenomena
RT HapMap Project
Human Genome Diversity Project
Population genetics+
CL 15.+

**GENETIC ENGINEERING**
DT 1974 (was GENETIC INTERVENTION, 1974-2001)
SN Directed modification of the gene complement of a living organism by such techniques as altering the DNA, substituting genetic material by means of a virus, transplanting whole nuclei, transplanting cell hybrids, etc. (MeSH)
BT Bioengineering
Genetic intervention
NT Biotechnology+
Chimeras
Gene therapy
Genetic enhancement
Genetically modified food
Genetically modified organisms+
Recombinant DNA research
RT Containment (*of biohazards*)
Genetic Techniques+

CL 15.1 (or 15.7 for biohazards of genetic research)

**GENETIC ENHANCEMENT**
DT 1996 (was GENE THERAPY or GENETIC INTERVENTION, 1974-1995)
SN The use of genetic technologies to augment or improve human or animal capacities or traits, rather than to treat disease
BT Enhancement technologies
Genetic engineering
RT Eugenics
Gene therapy
Gene transfer techniques
CL 15.+ and 4.5

**GENETIC EPIDEMIOLOGY**
DT 2008
SN The study of genetics as a phenomenon of defined populations by the criteria, methods, and objectives of epidemiology rather than of population genetics (Online Medical Dictionary, accessed 1/17/08)
EN Epidemiological genetics
BT Epidemiology
Genetics
RT Genetic research+
Population genetics+
CL 15.+

**GENETIC IDENTITY**
DT 1998
SN Personal identity as it is based on the individual's genotype
BT Psychosocial genetics
RT Self concept
CL 15.+

**GENETIC INFORMATION**
DT 1997
SN Genetic data, about an individual, family, or population group, derived from genetic research, tests, or family histories
BT Genetics
NT Genetic databases
Genetic privacy
Pedigree
RT Genetic counseling
Genetic research+
Genetic testing
Incidental findings
CL 15.+

**GENETIC INTERVENTION**
DT 1974
SN General term for the modification of inheritable characteristics of individuals or populations through various social mechanisms and/or genetic technologies
BT Genetics
NT Eugenics
Genetic counseling
Genetic engineering+

Genetic testing
Preimplantation diagnosis
Prenatal diagnosis+
RT Clinical genetics+
Genetic research+
Sex determination
Sex preselection
CL 15.+

**GENETIC MARKERS**
DT 2007
SN Specific DNA sequences that are associated with the variability of a genetic disorder or trait and that can potentially be used to determine the locus of the gene or genes associated with that trait or disorder
BT Genetic phenomena
CL 15.+

**GENETIC MATERIALS**
DT 1997
SN DNA and biological entities from which DNA can be extracted
BT Genetics
NT DNA
DNA sequences
Genes+
Genetic resources
Genome+
RT Genetic patents
CL 15.+

**GENETIC PATENTS**
DT 2007 (was PATENTS, 1974-2001)
SN Patents on life forms or genetic materials
UF Biopatents
BT Genetic research
Patents
RT Genetic materials+
CL 15.8

**GENETIC PHENOMENA**
DT 2007
SN Concepts related to genetic processes and structures
BT Genetics
NT Epigenetics
Evolution
Gene pool
Genetic determinism
Genetic diversity
Genetic markers
Genotype+
Mutation
Nuclear reprogramming
Phenotype
Single nucleotide polymorphisms
CL 15.+

**GENETIC PREDISPOSITION**
DT 1995
SN The presence of genetic factors indicating a higher-than-average risk, but not the certainty, that an individual will develop a disorder later in life
BT Genotype
RT Genetic disorders+
Genetic testing
CL 15.2 or 15.3

**GENETIC PRIVACY**
DT 2007 (was GENETIC INFORMATION, 1997-2001)
SN The protection of genetic information about an individual, family, or population group, from unauthorized disclosure (MeSH)
BT Genetic information
Privacy
RT Confidentiality+
DNA fingerprinting
Genetic databases
Genetic research+
Genetic testing
CL 15.+ and 8.4

**GENETIC RELATEDNESS TIES**
DT 2007
SN The psychological significance of being linked genetically to another person, for example, one's child
UF Genetic kinship ties
Genetic relationship ties
Kinship ties, genetic
BT Psychosocial genetics
CL (15.1 or 15.2) and/or 14.+

**GENETIC RESEARCH**
DT 1995
SN Broadly, investigation into the cause, transmission, amelioration, elimination, or enhancement of inherited disorders and/or traits
BT Biomedical research
Genetics
NT Genetic patents
HapMap Project
Human Genome Diversity Project
Human Genome Project
Recombinant DNA research
RT Genetic databases
Genetic epidemiology
Genetic information+
Genetic intervention+
Genetic privacy
Genome mapping+
Research subjects+
CL 15.1 (or 15.7 for biohazards of genetic research)

**GENETIC RESOURCES**
DT 2008

SN Genetic materials found in an ecosystem or a population group that are of actual or potential value in commerce or research; not used to index literature or database resources on genetics
BT Genetic materials
RT Benefit sharing
Biodiversity
CL 15.+

**GENETIC SERVICES**
DT 1997
SN Organized services to provide diagnosis, treatment, and prevention of genetic disorders
BT Clinical genetics
RT Gene therapy
Genetic counseling
Genetic testing
Preimplantation diagnosis
Prenatal diagnosis+
CL 15.2 or 15.3 or 15.4

**GENETIC TECHNIQUES**
DT 2007
SN Chromosomal, biochemical, intracellular, and other methods used in the study of genetics (MeSH)
BT Genetics
NT Cloning+
DNA fingerprinting
Gene transfer techniques
Genetic association studies+
Genome mapping+
Nuclear transfer techniques+
RT Genetic engineering+
Pedigree
CL 15.+

**GENETIC TESTING**
DT 1974
SN The use of, on a (smaller or larger) population group, of diagnostic procedures intended to identify either individuals affected by a genetic disorder or asymptomatic carriers of a particular genetic trait
UF Genetic screening
Predictive genetic testing (use GENETIC TESTING and (predictive or GENETIC PREDISPOSITION or LATE-ONSET DISORDERS+))
Public health-oriented genetic screening (use GENETIC TESTING and (MASS SCREENING or PUBLIC HEALTH))
Screening, genetic
Testing, genetic
BT Clinical genetics
Genetic intervention
RT DNA fingerprinting
Genetic counseling
Genetic information+
Genetic predisposition
Genetic privacy
Genetic services

Mandatory testing
Mass screening
Preimplantation diagnosis
Prenatal diagnosis+
CL 15.3

**GENETICALLY MODIFIED ANIMALS**
DT 2007 (was TRANSGENIC ANIMALS, 1990-2001)
SN Animals whose genomes have been altered by genetic engineering, or their offspring (MeSH)
UF GM animals
Transgenic animals
BT Genetically modified organisms
RT Genetically modified food
CL 15.1 and 22.3

**GENETICALLY MODIFIED FOOD**
DT 2007
SN Food derived from genetically modified organisms (MeSH)
UF GM food
BT Genetic engineering
RT Genetically modified animals
Genetically modified plants
CL 15.1 and 1.3.11

**GENETICALLY MODIFIED ORGANISMS**
DT 2007 (was TRANSGENIC ORGANISMS, 1997-2001)
SN Organisms whose genome has been changed by a genetic engineering technique (MeSH)
UF GMOs
Transgenic organisms
BT Genetic engineering
NT Genetically modified animals
Genetically modified plants
RT Genetically modified food
Transgenes
CL 15.1 and (1.3.11 or 22.3)

**GENETICALLY MODIFIED PLANTS**
DT 2007
SN Plants, or their progeny, whose genomes have been altered by genetic engineering (MeSH)
UF Genetically modified crops
Genetically modified seeds
GM plants
Transgenic plants
BT Genetically modified organisms
RT Genetically modified food
CL 15.1 and 1.3.11

**GENETICS**
DT 1974
SN The branch of science concerned with the means and consequences of transmission and generation of the components of biological inheritance (Stedman's Online Medical

Dictionary)
BT Biology
NT Behavioral genetics+
Clinical genetics+
Forensic genetics+
Genetic disorders+
Genetic epidemiology
Genetic information+
Genetic intervention+
Genetic materials+
Genetic phenomena+
Genetic research+
Genetic techniques+
Genomics+
Pharmacogenetics
Population genetics+
Psychiatric genetics
Psychosocial genetics+
Sociobiology
CL 15.+

**GENOME**
DT 2007
SN All of the DNA contained in an organism or a cell, which includes both the chromosomes within the nucleus and the DNA in mitochondria (National Human Genome Research Institute Talking Glossary, 2007)
BT Genetic materials
NT Human genome
RT DNA sequences
Genomics+
CL 15.+

**GENOME MAPPING**
DT 1989
SN The use of genetic techniques to produce a detailed overview of the gene locations within a genome
BT Genetic techniques
NT Human Genome Project
RT Genetic research+
CL 15.10

**GENOME-WIDE ASSOCIATION STUDIES**
DT 2008
SN Studies of genetic variation across the entire human genome that are designed to identify genetic associations with measurable traits (such as blood pressure or weight), or the presence of absence of a disease or condition (Genome-Wide Association Studies (GWAS) Web Site, accessed 2/14/08)
UF Whole-genome association studies
BT Genetic association studies
CL 15.+

**GENOMICS**
DT 2007
SN The systematic study of the complete DNA sequences (genomes) of organisms (MeSH)

BT Genetics
NT Human Genome Project
Nutrigenomics
Personal genomics
Proteomics
RT Genome+
Human Genome Project
CL 15.1

**GENOTYPE**
DT 2007
SN The genetic makeup, as distinguished from the physical appearance, of an organism
BT Genetic phenomena
NT Genetic carriers
Genetic predisposition
Haplotypes
RT Phenotype
CL 15.+

**HAPLOTYPES**
DT 2010
SN Sets of DNA variations, or polymorphisms, that tend to be inherited together (National Human Genome Research Institute Talking Glossary of Genetic Terms, accessed 1/2010)
BT Genotype
RT HapMap Project
CL 15.+

**HapMap PROJECT**
DT 2007
SN An international effort to identify and catalog genetic similarities and differences in human beings. Using the information in the HapMap, researchers will be able to find genes that affect health, disease, and individual responses to medications and environmental factors. The Project is a collaboration among scientists and funding agencies from Japan, the United Kingdom, Canada, China, Nigeria, and the United States (from the International HapMap Project webpage)
UF International HapMap Project
BT Genetic research
RT Genetic diversity
Haplotypes
Population genetics+
CL (15.1 or 15.10) and 13.1 and 21.1

**HEMOGLOBINOPATHIES**
DT 2010
SN A group of inherited disorders characterized by structural alterations within the hemoglobin molecule (MeSH)
BT Genetic disorders
NT Sickle cell anemia
Thalassemia
CL 15.+ and scattered

**HEMOPHILIA**
DT 1974
SN A group of inherited bleeding disorders in which the ability of blood to clot is impaired

(Medicine.net, accessed 11/25/08)
BT Genetic disorders
CL 15.+

**HUMAN GENOME**
DT 2007
SN The complete genetic complement contained in the DNA of a set of chromosomes in a human (MeSH, truncated)
BT Genome
CL 15.+

**HUMAN GENOME DIVERSITY PROJECT**
DT 2007 (was GENETIC DIVERSITY, 1998-2001)
SN An international research effort, under the auspices of the Human Genome Organization (HUGO), to collect biological samples from different population groups throughout the world, with the aim of building a representative database of human genetic diversity
UF HGDP
BT Genetic research
RT Genetic diversity
Human Genome Project
Population genetics+
CL 15.10 and 13.1 and 21.1

**HUMAN GENOME PROJECT**
DT 2007 (was GENOME MAPPING, 1989-2001)
SN An international research effort, completed in April 2003, to map and sequence all of the genes of the human genome
UF HGP
BT Genetic research
Genome mapping
Genomics
RT Human genome
Human Genome Diversity Project
CL 15.10

**HUNTINGTON DISEASE**
DT 1974
SN A familial disorder inherited as an autosomal dominant trait and characterized by the onset of progressive chorea and dementia in the fourth or fifth decade of life (MeSH, truncated)
BT Late-onset disorders
CL 15.+

**INDUCED PLURIPOTENT STEM CELLS**
DT 2009
SN Pluripotent stem cells that are artificially derived from non-pluripotent cells, typically adult somatic cells, through a variety of laboratory techniques
UF iPS cells
BT Pluripotent stem cells
RT Adult stem cells
Altered nuclear transfer
CL 18.7

**KLINEFELTER SYNDROME**

DT 2010
SN A chromosomal condition that affects male sexual development (Genetics Home Reference, accessed 1/20/10)
BT Chromosome abnormalities
CL 15.+

**LATE-ONSET DISORDERS**
DT 1995 (was GENETIC DISORDERS *or* HUNTINGTON'S DISEASE, 1974-1994)
UF Adult-onset genetic disorders
Late-onset genetic disorders
BT Genetic disorders
NT Huntington disease
CL 15.+

**MUTATION**
DT 2010
SN Any detectable and heritable alteration in the genetic material that causes a change in the genotype and that is transmitted to daughter cells and to succeeding generations (MeSH, truncated)
BT Genetic phenomena
CL 15.+

**NUCLEAR REPROGRAMMING**
DT 2009
SN The process that reverts cell nuclei of fully differentiated somatic cells to a pluripotent or totipotent state
BT Genetic phenomena
RT Nuclear transfer techniques+
CL 15.1 or 18.7

**NUCLEAR TRANSFER TECHNIQUES**
DT 2007
SN Methods of implanting a cell nucleus from a donor cell into an enucleated acceptor cell. Often the nucleus of a somatic cell is transferred into a recipient ovum or stem cells with the nucleus removed (adapted from MeSH)
BT Genetic techniques
NT Altered nuclear transfer
RT Cloning+
Nuclear reprogramming
Stem cells+
CL 14.5 or 15.1

**NUTRIGENOMICS**
DT 2008
SN The study of how different foods may interact with those having specific genotypes to increase or decrease the risk of common chronic diseases such as type-2 diabetes, obesity, heart disease, stroke, and certain cancers; nutrigenomics also seeks to provide a molecular understanding of how common chemicals in the diet affect health by altering the expression of genes and the structure of an individual's genome (MedicineNet.com, accessed 2/14/08)
UF Genomics, Nutritional
Nutrigenetics
BT Genomics
RT Nutrition

CL 15.1

**PEDIGREE**
DT 2007
SN A record of descent or ancestry, particularly of a particular condition or trait, indicating individual family members, their relationships, and their status with respect to the trait or condition (MeSH)
BT Genetic information
RT Genetic ancestry
Genetic techniques+
CL 15.+

**PERSONAL GENOMICS**
DT 2008
SN Analysis of the DNA of a particular individual's genome
UF Genomics, personal
BT Genomics
RT Personalized medicine
Pharmacogenetics
CL 15.1 or 15.10

**PHARMACOGENETICS**
DT 2007
SN A branch of genetics that deals with the genetic variability in individual responses to drugs and drug metabolism
UF Pharmacogenomics
BT Genetics
RT Genetic ancestry
Personal genomics
Toxicogenetics
CL 15.1 and 9.7

**PHENOTYPE**
DT 2007
SN The outward appearance of an organism, as determined by both its genetic makeup and environmental influences
BT Genetic phenomena
RT Genotype+
CL 15.+

**PHENYLKETONURIA**
DT 1974
SN A genetic disorder in which the body lacks the enzyme necessary to metabolize phenylalanine to tyrosine. Left untreated, the disorder can cause brain damage and progressive mental retardation as a result of the accumulation of phenylalanine and its breakdown products (American Heritage Dictionary of the English Language, 4th ed., 2006)
UF PKU
BT Genetic disorders
CL 15.+

**PLURIPOTENT STEM CELLS**
DT 2008
SN Cells that are able to develop into many different types of cells or tissues in the body or in

tissue culture
BT Stem cells
NT Induced pluripotent stem cells
CL 18.7

**POPULATION GENETICS**
DT 2000
SN The study of the genetic composition of populations
UF Genetics, population
BT Genetics
NT Genetic ancestry
RT Gene pool
Genetic diversity
Genetic epidemiology
HapMap Project
Human Genome Diversity Project
CL 15.1 and 13.1

**PREIMPLANTATION DIAGNOSIS**
DT 1991
SN Determination of the genetic makeup of an early human embryo prior to implantation
BT Clinical genetics
Diagnosis
Genetic intervention
RT Genetic services
Genetic testing
Prenatal diagnosis+
Sex predetermination
CL 15.2 and 14.4

**PRENATAL DIAGNOSIS**
DT 1974
SN Determination of fetal status prior to birth
UF Alpha-fetoprotein screening
Antenatal diagnosis
BT Clinical genetics
Diagnosis
Genetic intervention
NT Amniocentesis
Chorionic villi sampling
Sex determination
RT Fetal therapy
Genetic counseling
Genetic services
Genetic testing
Preimplantation diagnosis
CL 15.2

**PROTEOMICS**
DT 2008
SN The study of the expression, localizations, functions, and interactions of the full set of proteins encoded by a genome ((American Heritage Science Dictionary, 2005, truncated)
BT Genomics+
CL 15.+

**PSYCHIATRIC GENETICS**
DT 2009; *was* GENETICS *and* PSYCHIATRY
SN The study of the genetic causes and modes of inheritance that underlie the generally recognized mental illnesses (answers.com)
BT Genetics
RT Psychiatry
CL 15.1 and 17.1

**PSYCHOSOCIAL GENETICS**
DT 2007
SN The psychological and cultural impact of genetic factors on behaviors, attitudes, and beliefs; do not confuse with Behavioral Genetics, which is a branch of genetics that studies effects of genetics on behavior
UF Geneticization
BT Genetics
Psychology
NT Genetic discrimination
Genetic identity
Genetic relatedness ties
CL 15.+

**RECOMBINANT DNA RESEARCH**
DT 1978
SN Research using biologically active DNA that has been formed in the laboratory by the joining of segments of DNA from different sources
BT Genetic engineering
Genetic research
RT Containment of biohazards
CL 15.1 (or 15.7 for biohazards of recombinant DNA research)

**RNA**
DT 2008
SN A nucleic acid found in all living cells that plays many roles in the storage and transmission of genetic information, including the delivery of DNA's genetic message to the cytoplasm, where proteins are made
UF Ribonucleic acid
BT Genetic materials
CL 15.+

**SICKLE CELL ANEMIA**
DT 1974
SN A disease passed down through families in which red blood cells are an abnormal crescent shape (Millenium MedlinePlus Medical Encylopedia, accessed 12/4/08)
BT Hemoglobinopathies
CL 15.+ or scattered

**SINGLE NUCLEOTIDE POLYMORPHISMS**
DT 2010
SN Single nucleotide variations in a genetic sequence that occur at appreciable frequency in the population (MeSH); scientists are studying how single nucleotide polymorphisms, or SNPs (pronounced "snips"), in the human genome correlate with disease, drug response, and other phenotypes (National Human Genome Research Institute Talking Glossary of Genetic Terms, accessed 1/20/10)
UF Polymorphisms, single nucleotide

SNPs
BT Genetic phenomena
CL 15.+

**SOCIOBIOLOGY**
DT 1977
SN The study of biological influences on social behavior, based on the theory that such behavior is often genetically transmitted and subject to evolutionary processes (adapted from Amer Heritage Dictionary of the English Language, 4th ed, 2000)
BT Genetics
RT Behavioral genetics+
Evolution
CL 15.9

**STEM CELLS**
DT 2007
SN Cells with the ability to divide for indefinite periods in culture and to give rise to more specialized cells
BT Body parts and fluids
NT Adult stem cells
Embryonic stem cells
Pluripotent stem cells+
RT Stem cell transplantation
CL 18.7

**SYNTHETIC BIOLOGY**
DT 2008
SN The field at the interface of engineering and biology that involves designing and building systems from biological components (Nature Reviews Genetics: Glossary Terms, accessed 1/22/08)
UF Artificial life†
Life, synthetic†
Synthetic life†
BT Biology
Biotechnology
CL 1.3.9 and/or 15.1

**TAY SACHS DISEASE**
DT 1974
SN An autosomal recessive neurodegenerative disorder characterized by the onset in infancy of an exaggerated startle response, followed by paralysis, dementia and blindness (MeSH, truncated)
BT Genetic disorders
CL 15.+

**THALASSEMIA**
DT 1977
SN A group of inherited disorders characterized by reduced or absent amounts of hemoglobin, the oxygen-carrying protein inside the red blood cells (answers.com, accessed ll/25/08)
BT Hemoglobinopathies
CL 15.+

**TOXICOGENETICS**
DT 2009; *was* GENETICS *and* (HEALTH HAZARDS *or* TOXICITY), 1974-2001

SN The study of existing genetic knowledge, and the generation of new genetic data, to understand and thus avoid drug toxicity and adverse effects from toxic substances in the environment (MeSH)
UF Toxicogenomics
BT Genetics
RT Health hazards
Pharmacogenetics
Toxicity
CL 15.1 (often *with* 9.7 or 16.1)

**TRANSGENES**
DT 2010
SN Genes that have been introduced into an organism using gene transfer techniques (MeSH, truncated); in some circumstances these genes can then spread from the genetically modified organism into wild populations
BT Genes
RT Genetically modified organisms+
CL 15.+

**TRISOMY**
DT 2008
SN The possession of a third chromosome number at a particular location in the genome when two chromosomes would be the usual number; trisomy 21 is also called Down syndrome
BT Chromosome abnormalities
NT Down syndrome
CL 15.+

**TURNER SYNDROME**
DT 2010
SN A chromosomal condition that affects development in females (Genetics Home Reference, accessed 1/20/10)
BT Chromosome abnormalities
CL 15.+

**XYY KARYOTYPE**
DT 1975
SN A chromosome arrangement in males that is characterized by an extra Y chromosome
BT Chromosome abnormalities
RT Behavioral genetics+
CL 15.+

**GENETICS TREE FOR THE 2010 BIOETHICS THESAURUS**

- GENETICS
  - BEHAVIORAL GENETICS
    - GENETIC DETERMINISM
  - CLINICAL GENETICS
    - GENE THERAPY
    - GENETIC COUNSELING
    - GENETIC SERVICES
    - GENETIC TESTING
    - PREIMPLANTATION DIAGNOSIS
    - PRENATAL DIAGNOSIS
  - FORENSIC GENETICS
    - DNA FINGERPRINTING
  - GENETIC DISORDERS
    - CANAVAN DISEASE
    - CHROMOSOME ABNORMALITIES
      - FRAGILE X SYNDROME
      - KLINEFELTER SYNDROME
      - TRISOMY
        - DOWN SYNDROME
      - TURNER SYNDROME
      - XYY KARYOTYPE
    - CYSTIC FIBROSIS
    - DUCHENNE MUSCULAR DYSTROPHY
    - GAUCHER DISEASE
    - HEMOGLOBINOPATHIES
      - SICKLE CELL ANEMIA
      - THALASSEMIA
    - HEMOPHILIA
    - KLINEFELTER SYNDROME
    - LATE-ONSET DISORDERS
      - HUNTINGTON DISEASE
    - PHENYLKETONURIA
    - TAY SACHS DISEASE
  - GENETIC EPIDEMIOLOGY
  - GENETIC INFORMATION
    - GENETIC DATABASES
    - GENETIC PRIVACY
    - PEDIGREE
  - GENETIC INTERVENTION
    - EUGENICS
    - GENETIC COUNSELING
    - GENETIC ENGINEERING
      - BIOTECHNOLOGY
        - SYNTHETIC BIOLOGY
      - CHIMERAS
      - GENE THERAPY
      - GENETIC ENHANCEMENT
      - GENETICALLY MODIFIED FOOD
      - GENETICALLY MODIFIED ORGANISMS
        - GENETICALLY MODIFIED ANIMALS
        - GENETICALLY MODIFIED PLANTS
      - RECOMBINANT DNA RESEARCH

- GENETIC TESTING
- PREIMPLANTATION DIAGNOSIS
- PRENATAL DIAGNOSIS

GENETIC MATERIALS
- DNA
- DNA SEQUENCES
- GENES
  - APO-E GENES
  - BRCA1 GENES
  - BRCA2 GENES
  - TRANSGENES
- GENETIC RESOURCES
- GENOME
  - HUMAN GENOME
- RNA

GENETIC PHENOMENA
- EPIGENETICS
- EVOLUTION
- GENE POOL
- GENETIC DETERMINISM
- GENETIC DIVERSITY
- GENETIC MARKERS
- GENOTYPE
  - GENETIC CARRIERS
  - GENETIC PREDISPOSITION
  - HAPLOTYPES
- MUTATION
- NUCLEAR REPROGRAMMING
- PHENOTYPE
- SINGLE NUCLEOTIDE POLYMORPHISMS

GENETIC RESEARCH
- GENETIC PATENTS
- HapMap PROJECT
- HUMAN GENOME DIVERSITY PROJECT
- HUMAN GENOME PROJECT
- RECOMBINANT DNA RESEARCH

GENETIC TECHNIQUES
- CLONING
  - ANIMAL CLONING
- DNA FINGERPRINTING
- GENE TRANSFER TECHNIQUES
- GENETIC ASSOCIATION STUDIES
  - GENOME-WIDE ASSOCIATION STUDIES
- GENOME MAPPING
- NUCLEAR TRANSFER TECHNIQUES
  - ALTERED NUCLEAR TRANSFER

GENOMICS
- HUMAN GENOME PROJECT
- NUTRIGENOMICS
- PERSONAL GENOMICS
- PROTEOMICS

PHARMACOGENETICS

POPULATION GENETICS
- GENETIC ANCESTRY

PROTEOMICS
PSYCHIATRIC GENETICS
PSYCHOSOCIAL GENETICS
    GENETIC DISCRIMINATION
    GENETIC IDENTITY
    GENETIC RELATEDNESS TIES
SOCIOBIOLOGY
STEM CELLS
    ADULT STEM CELLS
    EMBRYONIC STEM CELLS
    PLURIPOTENT STEM CELLS
        INDUCED PLURIPOTENT STEM CELLS
TOXICOGENETICS

## APPENDIX:
## EXAMPLES OF GENETICS-RELATED KEYWORD IDENTIFIERS, 2010

Advisory Group on Human Gene Patents and Genetic Testing (Australia)
Alberta Eugenics Board
American Society of Medical Genetics
Asilomar Conference
BiDil
BioBank Japan Project
California Institute for Regenerative Medicine
Celera Genomics
Church, George
Collins, Francis
Convention on Biological Diversity
deCode Genetics
deCODEMe
Diamond v. Chakrabarty
Empire State Stem Cell Board
Estonian Genome Project
EuroBioBank
European Society of Human Genetics
Genetic Information Nondiscrimination Act (GINA)
Geron Corp.
Human Genetics Advisory Committee (Australia)
Human Genetics Commission (Great Britain)
Human Genome Organization (HUGO)
International Declaration on Human Genetic Data
International Society of Nurses in Genetics
Monsanto Company
Myriad Genetics Inc.
National Human Genome Research Institute
National Institute for Human Genome Research
National Society of Genetic Counselors
Navigenics
NCHGR Program on Ethical, Legal, and Social Implications (ELSI)
NHGRI Program on Ethical, Legal, and Social Implications (ELSI)
NIH-DOE Working Group on Ethical, Legal, and Social Implications (ELSI)
1,000 Genomes Project
Personal Genome Project
Public Population Project in Genomics
Recombinant DNA Advisory Committee
Schmeister v. Monsanto Company
Secretary's Advisory Committee on Genetics, Health, and Society [SACGHS]
Statement on Human Genomic Databases (HUGO Ethics Committee)
Taiwan Biobank Project
Targeted Genetics Corp.
23andMe
UK Biobank
Universal Declaration on the Human Genome and Human Rights
Venter, J. Craig