Glossary of Terms

**Genetics** is a term that refers to the study of genes and their role in inheritance – the way certain traits are passed down from one generation to another.

**Genomics** is the study of all of a person’s genetic material, the interactions of that genetic material, the interactions of that genetic material with the environment, and the subsequent phenotypic changes.

**Genetics** uses information from a few genes to explain a disease or condition, whereas **genomics** examines all of the genetic information to determine biological markers predisposing an individual to disease.

**Autosomal Dominant**
Autosomal dominance is a pattern of inheritance characteristic of some genetic diseases. "Autosomal" means that the gene in question is located on one of the numbered, or non-sex, chromosomes. "Dominant" means that a single copy of the disease-associated mutation is enough to cause the disease. This is in contrast to a recessive disorder, where two copies of the mutation are needed to cause the disease. Huntington’s disease is a common example of an autosomal dominant genetic disorder.

**Bioinformatics**
Bioinformatics is a subdiscipline of biology and computer science concerned with the acquisition, storage, analysis, and dissemination of biological data, most often DNA and amino acid sequences. Bioinformatics uses computer programs for a variety of applications, including determining gene and protein functions, establishing evolutionary relationships, and predicting the three-dimensional shapes of proteins.

**Carrier**
A carrier is an individual who carries and is capable of passing on a genetic mutation associated with a disease and may or may not display disease symptoms. Carriers are associated with diseases inherited as recessive traits. In order to have the disease, an individual must have inherited mutated alleles from both parents. An individual having one normal allele and one mutated allele does not have the disease. Two carriers may produce children with the disease.

**Carrier Screening**
Carrier screening is a type of genetic testing performed on people who display no symptoms for a genetic disorder but may be at risk for passing it on to their children. A carrier for a genetic disorder has inherited one normal and one abnormal allele for a gene associated with the disorder. A child must inherit two abnormal alleles in order for symptoms to appear. Prospective parents with a family history of a genetic disorder are candidates for carrier screening.

**Copy Number Variation (CNV)**
A copy number variation (CNV) is when the number of copies of a particular gene varies from one individual to the next. CNV is a type of structural variation where you have a stretch of DNA, which is duplicated in some people, and sometimes even triplicated or quadruplicated. Sometimes those CNVs include genes, maybe several genes, which may mean that this person has four copies of that gene instead of the usual two, and somebody else has three, and somebody else has five. The extent
to which copy number variation contributes to human disease is not yet known. CNVs that do not cause disease are benign variants. It has long been recognized that some cancers are associated with elevated copy numbers of particular genes. Microdeletion CNVs are more commonly associated with disease.

**DNA Sequencing**

DNA sequencing is a laboratory technique used to determine the exact sequence of bases (A, C, G, and T) in a DNA molecule. The DNA base sequence carries the information a cell needs to assemble protein and RNA molecules. DNA sequence information is important to scientists investigating the functions of genes. The technology of DNA sequencing was made faster and less expensive as a part of the Human Genome Project.

**Epigenetics**

The processes that regulate gene expression in a given cell leading to its cellular phenotype. (Conrad Waddington 1942)

Epigenetics is an emerging field of science that studies heritable changes caused by the activation and deactivation of genes without any change in the underlying DNA sequence of the organism. It is the study of changes in gene function that are heritable and that are not attributed to alterations in the DNA sequence. Epigenetics can influence disease penetrance and variability among individuals.

**Exome Sequencing**

Exome sequencing is a technique that is used to examine all of the protein-coding regions of the genome.

**Family History**

A family history is a record of medical information about an individual and their biological family. Human genetic data is becoming more prevalent and easy to obtain. Increasingly, this data is being used to identify individuals who are at increased risk for developing genetic disorders that run in families.

**FISH (Fluorescence In Situ Hybridization)**

Fluorescence in situ hybridization (FISH) is a laboratory technique for detecting and locating a specific DNA sequence on a chromosome. In this approach, a fluorescent dye is attached to a purified piece of DNA, and then that DNA is incubated with the full set of chromosomes from the originating genome, which have been attached to a glass microscope slide. The fluorescently labeled DNA finds its matching segment on one of the chromosomes, where it sticks. By looking at the chromosomes under a microscope, a researcher can find the region where the DNA is bound because of the fluorescent dye attached to it. This information thus reveals the location of that piece of DNA in the starting genome. FISH can be used to detect trisomies, William’s syndrome, and DiGeorge syndrome among other conditions.

**Genetic Counseling**

Genetic counseling is the process of helping people understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease. This process integrates the following:

- Interpretation of family and medical histories to assess the chance of disease occurrence.
- Education about inheritance, testing, management, prevention, resources, and research.
- Counseling to promote informed choices and adaptation to the risk or condition.
Genetic counseling is the professional interaction between a healthcare provider with specialized knowledge of genetics and an individual or family. The genetic counselor determines whether a condition in the family may be genetic and estimates the chances that another relative may be affected. Genetic counselors also offer and interpret genetic tests that may help to estimate risk of disease. The genetic counselor conveys information in an effort to address concerns of the client and provides psychological counseling to help families adapt to their condition or risk.

Genetic counseling is also something that primary care providers should be incorporating as part of their routine appointments with patients with genetic conditions or at risk for developing a genetic condition.

**Genetic/Genomic Literacy**
The degree to which individuals have the capacity to understand and utilize genetics knowledge as it relates to and impacts the lives of individuals.

**Genetic Tests**
Genetic testing is the use of a laboratory test to look for genetic variations associated with a disease. The results of a genetic test can be used to confirm or rule out a suspected genetic disease or to determine the likelihood of a person passing on a mutation to their offspring.

**Genetics**
Genetics is the study of inherited biological variation. Genetics is a term that refers to the study of genes and their role in inheritance – the way certain traits or conditions are passed down from one generation to another. Genetics involves scientific studies of single genes and their effects. Genes (units of heredity) carry the instructions for making proteins, which direct the activities of cells and functions of the body. Genes influence traits such as hair and eye color as well as health and disease development. Genetics determines much (but not all) of a person’s appearance and health status, but environmental differences also play a part.

**Genome**
The genome is the entire set of genetic instructions found in a cell. In humans, the genome consists of 23 pairs of chromosomes, found in the nucleus, as well as a small chromosome found in the cell’s mitochondria.

**Genomics**
Genomics refers to the study of the entire genome of an organism. Genomics is the study of all of a person’s genes including interactions of those genes with each other and the person’s environment. Genomics involves the scientific study of complex diseases such as heart disease, asthma, diabetes and cancer because they are caused more by a combination of genetic and environmental factors. Genomics is offering new possibilities for therapies and treatment of some diseases, as well as new diagnostic methods. The major tools and methods related to genomics studies are bioinformatics, genetic analysis, measurement of gene expression, and determination of gene function.

**Karyotype**
A karyotype is an individual’s collection of chromosomes. The term also refers to a laboratory technique that produces an image of an individual’s chromosomes. The karyotype is used to look for abnormal numbers or structures of chromosomes.
Microarray
Microarray technology is a developing technology used to study the expression of many genes at once. It involves placing thousands of gene sequences in known locations on a glass slide called a gene chip. A sample containing DNA or RNA is placed in contact with the gene chip. Complementary base pairing between the sample and the gene sequences on the chip produces light that is measured. Areas on the chip producing light identify genes that are expressed in the sample.

Minor Anomalies
Definition of minor anomalies from p. 895, Jones et al. “unusual morphologic features that are of no serious medical or cosmetic consequence to the patient...that may serve as indicators of altered morphogenesis in a general sense or may constitute valuable clues in the diagnosis of a specific pattern of malformation.

Newborn Screening
Newborn screening is testing performed on all babies to detect a wide variety of disorders that are treatable, or in certain cases preventable, with appropriate medical care that is provided essentially right away. Typically, testing is performed on a blood sample obtained from a heel prick when the baby is two or three days old. In the United States, newborn screening is mandatory for several different genetic disorders, though the exact set of required tests differs from state to state.

Personalized Medicine
Personalized medicine is an emerging practice of medicine that uses an individual’s genetic profile to guide decisions made in regard to the prevention, diagnosis, and treatment of disease. Knowledge of a patient’s genetic profile can help doctors select the proper medication or therapy and administer it using the proper dose or regimen. Personalized medicine is being advanced through data from the Human Genome Project.

Pedigree
A pedigree is a genetic representation of a family tree that diagrams the inheritance of a trait or disease through several generations. The pedigree shows the relationships between family members and indicates which individuals express or silently carry the trait in question.

Pharmacogenomics
Pharmacogenomics is a branch of pharmacology concerned with using DNA and amino acid sequence data to inform drug development and testing. An important application of pharmacogenomics is correlating individual genetic variation with drug responses.

Recessive
Recessive refers to a type of allele which will not be manifested in an individual unless both of the individual’s copies of that gene have that particular genotype. In the case of a recessive allele, the individual will show the trait which corresponds to that genotype only if both alleles are the same and have that particular recessive characteristic. This results in differences between individuals such as eye color or hair color, but it can also refer to a disease. For instance, in cystic fibrosis, which is a very common Mendelian disorder, the disease exists only when there is a malfunction of both genes that correspond to cystic fibrosis. If there is only one mutation, then that recessive mutation can be compensated for by the normal allele. However, when the function of both are lost, then the disease manifests itself as a recessive disease where there is a loss of function and therefore observable disease.
Sex Linked
Sex linked is a trait in which a gene is located on a sex chromosome. In humans, the term generally refers to traits that are influenced by genes on the X chromosome. This is because the X chromosome is large and contains many more genes than the smaller Y chromosome. In a sex-linked disease, it is usually males who are affected because they have a single copy of X chromosome that carries the mutation. In females, the effect of the mutation may be masked by the second healthy copy of the X chromosome.

Single Nucleotide Polymorphism (SNP)
A Single Nucleotide Polymorphism (SNP) is a base site that differs among individuals in a population. Analysis of SNPs in populations – for example, comparing the frequencies of selected SNPs between affected and unaffected persons – can provide clues to genetic variants associated with disease, including common diseases, and nutritional status. Sometimes the SNP is itself associated with the disease of interest, but often the SNP is only a nearby marker for the actual gene that affects susceptibility to the disease.

Southern Blot
Southern blotting is a laboratory technique used to detect a specific DNA sequence in a blood or tissue sample. A restriction enzyme is used to cut a sample of DNA into fragments that are separated using gel electrophoresis. The DNA fragments are transferred out of the gel to the surface of a membrane. The membrane is exposed to a DNA probe labeled with a radioactive or chemical tag. If the probe binds to the membrane, then the probe sequence is present in the sample.

Variant of Unknown Significance (VUS)
An alteration in the normal sequence of a gene, the significance of which is unclear until further study of the genotype and corresponding phenotype in a sufficiently large population; complete gene sequencing often identifies numerous (sometimes hundreds) allelic variants for a given gene.

Western Blot
Western blotting is a laboratory technique used to detect a specific protein in a blood or tissue sample. The method involves using gel electrophoresis to separate the sample's proteins. The separated proteins are transferred out of the gel to the surface of a membrane. The membrane is exposed to an antibody specific to the target protein. Binding of the antibody is detected using a radioactive or chemical tag. A western blot is sometimes used to diagnose disease.

Whole Genome Sequencing
Whole genome sequencing is a laboratory technique used to sequence the entire DNA sequence of an individual.