GLOSSARY OF TERMS:

· **acral-lentiginous melanoma -** An irregular, enlarging black flat spot (macule) with a prolonged noninvasive stage, occurring chiefly on the palms and soles; it is the most common type of melanoma in nonwhites.

· **Active transport -** The movement of molecules across a cell membrane in the direction against their concentration gradient, i.e. moving from a low concentration to a high concentration.

· **Acute Lymphoblastic Leukemia (ALL) -** An acute form of leukemia, or cancer of the white blood cells, characterized by the overproduction of cancerous, immature white blood cells—known as lymphoblasts.

· **Adenocarcinoma -** Neoplasia of epithelial tissue that has glandular origin, glandular characteristics, or both.

· **Adjuvant therapy -** Treatment that is given in addition to the primary, main or initial treatment.

· **Alpha-fetoprotein (AFP) -** A fetal glycoprotein excreted into the amniotic fluid that reaches abnormally high concentration in amniotic fluid (and maternal serum) when the fetus has certain abnormalities, especially an open neural tube defect.

· **Alzheimer's disease -** An irreversible, progressive brain disease that slowly destroys memory and thinking skills, and eventually even the ability to carry out the simplest tasks.

· **Amniocentesis -** A medical procedure used in prenatal diagnosis in which a small amount of amniotic fluid, which contains fetal cells, issampled from the amnion or amniotic sac surrounding a developing fetus, and examined for genetic abnormalities.

· **Aneuploidy -** Any chromosome number that is not an exact multiple of the haploid number. The common forms of aneuploidy in humans are trisomy (the presence of an extra chromosome) and monosomy (the absence of a single chromosome).

· **Antigen -** In immunology, an antigen, or antibody generator, is any substance which provokes an adaptive immune response. An

antigen is often foreign or toxic to the body which, once in the body, attracts and is bound to a respective and specific antibody.

· **Autosomal dominant -** A pattern of inheritance in which one copy of an autosomal gene must be abnormal for a genetic condition or disease to occur. An autosomal gene is a gene that is located on one of the autosomes or non-sex chromosomes.

· **Autosomal recessive -** A pattern of inheritance in which both copies of an autosomal gene must be abnormal for a genetic condition or disease to occur. An autosomal gene is a gene that is located on one of the autosomes or non-sex chromosomes.

· **Autosome -** Any nuclear chromosome other than the sex chromosomes; in humans there are 22 pairs of autosomes in a normal somatic cell.

· **Balanced translocation -** Translocation is transfer of a fragment of one chromosome to another chromosome. If two nonhomologous chromosomes exchange pieces, the translocation is reciprocal. A reciprocal translocation is referred to as balanced translocation, if there is an even exchange of material with no genetic information extra or missing.

· **Base Pair -** A pair of complementary nucleotide bases, as in double-stranded DNA. Used as the unit of measurement of the length of a DNA sequence.

· **BRCA 1 Gene –** Breast Cancer 1 gene**,** a human tumor suppressor gene found in all humans.

· **BRCA 2 Gene –** Breast Cancer 2 gene, a human tumor suppressor gene found in all humans.

· **Carrier -** An individual heterozygous for a particular mutant allele. The term is used for heterozygotes for autosomal recessive alleles, for females heterozygous for X-linked alleles, or, less commonly, for an individual heterozygous for an autosomal dominant allele but not expressing it (e.g., a heterozygote for a Huntington disease allele in the presymptomatic stage).

· **Cell line -** Cells grown in tissue culture and representing generations of a primary culture.

· **Centromere -** The primary constriction on the chromosome, a region at which the sister chromatids are held together and at which the kinetochore is formed. Required for normal segregation in mitosis and meiosis.

· **Chemotherapy -** The treatment of cancer using specific chemical agents or drugs that are selectively destructive to malignant cells and tissues.

· **Chorionic villus sampling (CVS) -** A procedure used for prenatal diagnosis at 8 to 10 weeks' gestation. Fetal tissue for analysis is withdrawn from the villous area of the chorion either transcervically or transabdominally, under ultrasonographic guidance.

· **Chromatin -** The complex of DNA and proteins of which chromosomes are composed.

· **Chromosome -** One of the threadlike structures in the cell nucleus; consists of chromatin and carries genetic information (DNA).

· **Clonal adaptation -** Cells undergo genetic or epigenetic changes in response to therapy.

· **Clonal selection -** Hererogeneity within the tumor leading to selection of resistant sub-clones.

· **Coding region -** The coding region of a gene, also known as the coding sequence or CDS, is that portion of a gene's DNA or RNA, composed of exons, that codes for protein.

· **Colitis -** Inflammation of the colon.

· **Congenital abnormalities -** A condition existing at birth and often before birth, or that develops during the first month of life (neonataldisease), regardless of causation.

· **Copy Number Variation (CNV) -** A variation in DNA sequence defined by the presence or absence of a segment of DNA, ranging from 200 bp to 2 Mb. Copy number variants may also have alleles that are

tandem duplications of two, three, four, or more copies of a DNA segment. If a variant has an allele frequency >1%, it is referred to as a copy number polymorphism (CNP).

· **Cultured -** The propagation of microorganisms or of living tissue cells in media conducive to their growth.

· **Cystic Fibrosis -** Cystic fibrosis is a disease passed down through families that causes thick, sticky mucus to build up in the lungs, digestive tract, and other areas of the body. The disorder is caused by mutations in the cystic fibrosis conductance regulator gene (CFTR), located on chromosome 7.

· **Cystic Hygroma -** An accumulation of fluid in a sac, cyst, or bursa.

· **Cytogenetics -** The study of chromosomes.

· **Cytopenic -** A disorder in which the production of one or more blood cell types ceases or is greatly reduced.

· **Deletion -** The loss of a sequence of DNA from a chromosome. The deleted DNA may be of any length, from a single base to a large part of a chromosome.

· **DiGeorge syndrome -** A congenital disorder characterized by severe immunodeficiency and structural abnormalities, including hypertelorism; notched, low-set ears; small mouth; downward slanting eyes; cardiovascular defects; and absence of the thymus and parathyroid glands. DiGeorge syndrome is caused by a 1.5- to 3.0-Mb hemizygous deletion of chromosome 22q11.2. Haploinsufficiency of the TBX1 gene in particular is responsible for most of the physical malformations.

· **Diploid -** The number of chromosomes in a normal somatic cell; in humans, the diploid number is 46 chromosomes (22 pairs of autosomes and two sex chromosomes).

· **Direct to consumer marketing (DTC) -** This form of advertising is directed toward patients, rather than healthcare professionals.

· **DNA (deoxyribonucleic acid) -** The molecule that encodes the genes responsible for the structure and function of living organisms and allows the transmission of genetic information from generation to generation.

· **DNA probes -** A radioactive or chemiluminescent DNA sequence used to detect the presence of a DNA complementary sequence.

· **DNA replication -** DNA replication is the process of producing two identical replicas from one original DNA molecule. This biological process occurs in all living organisms and is the basis for biological inheritance.

· **Double helix -** In molecular biology, the term double helix refers to the structure formed by double-stranded molecules of nucleic acids such as DNA.

· **Eggs -** The female reproductive cell which, after fertilization, becomes a zygote that develops into a new member of the same species.

· **Enzymes -** large biological molecules responsible for the thousands of metabolic processes that sustain life.

· **Epigenomics -** The study of the complete set of epigenetic modifications on the genetic material of a cell, known as the epigenome.

· **Ethos -** The distinguishing character, sentiment, moral nature, or guiding beliefs of a person, group, or institution.

· **Euploid -** Any chromosome number that is an exact multiple of the number of chromosomes in a haploid gamete.

· **Exome -** The portions of a gene or genome that code information for protein synthesis; the exons in the human genome.

· **False-Positive -** A positive test result in a subject that does not possess the attribute for which the test is being conducted.

· **Fatty acid synthase -** Fatty acid synthase is a multi-enzyme protein that catalyzes fatty acid synthesis.

· **Fixative -** An agent used in preserving a histological or pathological specimen so as to maintain the normal structure of its constituent elements.

· **Fluorescence In-situ Hybridization (FISH) -** Technique used to detect small deletions and duplications and rearrangements in chromosomes.

· **Fluorescent microscope -** A microscope fitted with a source of ultraviolet radiation to aid in the detection and examination of fluorescent specimens.

· **Food and Drug Administration (FDA) -** An agency of the United States Department of Health and Human Services, one of the United States federal executive departments. The FDA is responsible for protecting and promoting public health through the regulation and supervision offood safety, tobacco products, dietary supplements, prescription and over-the-counter pharmaceutical drugs (medications), vaccines,biopharmaceuticals, blood transfusions, medical devices, electromagnetic radiation emitting devices (ERED), cosmetics and veterinary products.

· **Fragile-X -** An X chromosome with a fragile site near the end of the long arm, resulting in the appearance of an almost detached fragment; demonstrated only under special culture conditions; frequently associated with X-linked mental retardation.

· **G-Banding -** A chromosome-staining technique used in cytogenetics to identify individual chromosomes, which produces characteristic bands.

· **Gamete -** A reproductive cell (ovum or sperm) with a haploid chromosome number.

· **Gene expression -** Conversion of the information encoded in a gene first into messenger RNA and then to a protein.

· **Gene therapy (gene transfer therapy) -** Treatment of a disease by introduction of DNA sequences that will have a therapeutic benefit.

· **Genes -** A hereditary unit; in molecular terms, a sequence of chromosomal DNA that is required for the production of a functional product.

· **Genetic counseling -** The provision of information and assistance to affected individuals or family members at risk of a disorder that may be genetic, concerning the consequences of the disorder, the probability of developing or transmitting it, and the ways in which it may be prevented or ameliorated.

· **Genotype -** The genetic constitution of an individual, as distinguished from the phenotype.

· **Germ cell -** Any biological cell that gives rise to the gametes of an organism that reproduces sexually. In many animals, the germ cells originate near the gut of an embryo and migrate to the developing gonads.

· **Growth media (or culture media) -** A liquid or gelatinous substance containing nutrients in which microorganisms or tissues are cultivated for scientific purposes.

· **Haploid -** The number of chromosomes in a normal gamete; in humans the haploid number is 23.

· **Hematologic malignancy -** Types of cancer that affects blood, bone marrow, and lymph nodes.

· **HER2/neu Gene -** HER2 is a member of the epidermal growth factor receptor (EGFR/ERBB) family. Amplification or overexpression of thisoncogene has been shown to play an important role in the development and progression of certain aggressive types of breast cancer.

· **Hernia -** is a general term used to describe a bulge or protrusion of an organ through the structure or muscle that usually contains it.

· **Histones -** Proteins associated with DNA in the chromosomes that are rich in basic amino acids (lysine or arginine) and virtually invariant throughout eukaryote evolution.

· **Human Genome Project -** A major research project, international in scope, that took place in the years 1990-2003 and resulted in the sequencing of a representative human genome..

· **Huntington's Disease -** A neurodegenerative genetic disorder that affects muscle coordination and leads to cognitive decline andpsychiatric problems. It typically becomes noticeable in mid-adult life. Huntington disease is caused by an expanded trinucleotide repeat (CAG)n, encoding glutamine, in the gene encoding huntingtin (HTT) on chromosome 4p16.3.

· **Hybridization -** In molecular biology, the bonding of two complementary single-stranded nucleic acid molecules according to the rules of base pairing.

· **Hypotonic -** Of or designating a solution of lower osmotic pressure than another.

· **Hysterectomy -** The surgical removal of the uterus.

· **Immunotherapy -** Treatment of disease by inducing, enhancing, or suppressing an immune response.

· **Incidental findings –** Genetic test results that were not anticipated.

· **Incubator -** An apparatus in which environmental conditions, such as temperature and humidity, can be controlled, often used for growing bacterial cultures, hatching eggs artificially, or providing suitable conditions for a chemical or biological reaction.

· **Insertion -** A chromosomal abnormality in which a DNA segment from one chromosome is inserted into another chromosome.

· **In-situ -** 1. In the natural or usual place; 2. Describing a cancer that has not metastasized or invaded neighboring tissues, such as carcinoma in situ.

· **Interphase -** The stage of the cell cycle between two successive mitoses.

· **Invasive Cancer-** Cancer that has spread beyond the layer of tissue in which it developed and is growing into surrounding, healthy tissues. Also called infiltrating cancer.

· **Karyotyping -** A laboratory test used to study an individual's chromosome make-up (number and structure of chromosomes). Chromosomes are separated from cells, stained, and arranged in order from longest to shortest.

· **KRAS Gene -** GTPase KRas also known as V-Ki-ras2 Kirsten rat sarcoma viral oncogene homolog and KRAS, is a protein that in humans is encoded by the KRAS gene.

· **Lesion -** Any pathological or traumatic discontinuity of tissue or loss of function of a part.

· **LOD Score -** A statistical method that tests genetic marker data in families to determine whether two loci (the sites of genes) are linked (likely to lie near each other on a chromosome) and are therefore likely to be inherited together. LOD stands for logarithm of the odds (to the base 10). By convention, a LOD score of 3 or more (odds are 1000: 1 or more in favor) is generally taken as proof of linkage and a LOD score of -2 (100:1 against), as proof that the loci are unlinked.

· **Lymph nodes -** Small, bean-shaped masses of tissue scattered along the lymphatic system that act as filters and immune monitors, removing fluids, bacteria, or cancer cells that travel through the lymph system.

· **Monogenic trait -** An inherited disease controlled by a single pair of genes.

· **Mast cell -** A cell found in connective tissue that releases substances such as heparin and histamine in response to injury or inflammation of bodily tissues.

· **Matabalomics -** The scientific study of chemical processes involving metabolites. Specifically, metabolomics is the "systematic

study of the unique chemical fingerprints that specific cellular processes leave behind", the study of their small-molecule metabolite profiles.

· **Melanoma -** A dark-pigmented, malignant, frequently widely metastasizing tumor arising from a melanocyte and occurring most commonly in the skin.

· **Mendelian disorder -** A genetic disease showing a mendelian pattern of inheritance, caused by a single mutation in the structure of DNA, which causes a single basic defect with pathologic consequences.

· **Meta-analysis -** Methods that focus on contrasting and combining results from different studies, in the hope of identifying patterns among study results, sources of disagreement among those results, or other interesting relationships that may come to light in the context of multiple studies.

· **Metabolites -** The intermediates and products of metabolism.

· **Metaphase -** The stage of mitosis or meiosis in which the chromosomes have reached their maximal condensation and are lined up on the equatorial plane of the cell, attached to the spindle fibers. This is the stage at which chromosomes are most easily examined.

· **Metastatic -** The term used to describe cancer that has spread from the primary area of the body where it started to another area of the body.

· **Microarray (Chromosomal) -** Miniaturized wafer ("chip") made of glass, plastic, or silicon onto which a large number of different nucleic acids have been individually spotted.

· **Microbiomics -** The scientific study of the microbiome.

· **Microdeletions -** A mutation (a genetic aberration) in which a part of a chromosome or a sequence of DNA is missing.

· **Monosomy -** A chromosome constitution in which one member of a chromosome pair is missing, as in 45,X Turner syndrome.

· **Morphology** - Is a branch of biology dealing with the study of the form and structure of organisms and their specific structural features.

· **Mosaicism -** A condition that occurs post-fertilization and in which there are two or more cell lines with different genetic or chromosomal constitutions within a single individual or tissue. Confined placental mosaicism (CPM) represents a discrepancy between the chromosomal makeup of the cells in the placenta and the cells in the baby.

· **Mucosa -** The mucous membrane, or the thin layer which lines body cavities and passages.

· **Mutation -** Any permanent heritable change in the sequence of genomic DNA**.**

· **Myometrium -** The myometrium is the middle layer of the uterine wall, consisting mainly of uterine smooth muscle cells, but also of supporting stromal and vascular tissue. Its main function is to induce uterine contractions.

· **Myopathy -** A muscular disease in which the muscle fibers do not function for any one of many reasons, resulting in muscular weakness.

· **Neutraceuticals -** The term is applied to products that range from isolated nutrients, dietary supplements and herbal products, specific diets and processed foods such as cereals, soups, and beverages.

· **Niemann-Pick -** An inherited disorder of lipid metabolism characterized by gastrointestinal disturbances and enlargement and abnormalities of blood-forming organs; it occurs primarily in infants of eastern European Jewish descent and it leads to early death. It is caused by an inherited deficiency of acid sphingomyelinase activity.

· **Non-coding region -** Or Antisense strand of DNA, is the noncoding DNA strand, which is complementary to mRNA and serves as the template for RNA synthesis. Also called the transcribed strand.

· **Non-invasive -** In cancer, it describes disease that has not spread outside the tissue in which it began.

· **Noonan syndrome -** Is a genetic disorder that prevents normal development in various parts of the body. A person can be affected by Noonan syndrome in various ways. These include unusual facial characteristics, short stature, heart defects, other physical problems and possible developmental delays. Noonan syndrome-1 is caused by heterozygous mutation in the PTPN11 gene on chromosome 12q24.1.

· **Nuchal fold -** Ultrasonographic measurement of skin thickness on the back of the fetal neck; measured from 15-22 weeks. Increased thickness indicates increased fetal risk for aneuploidy and some non-chromosomal disorders.

· **Nucleotide -** A molecule composed of a nitrogenous base, a 5-carbon sugar, and a phosphate group. A nucleic acid is a polymer of many nucleotides.

· **Nucleus -** A membrane-enclosed organelle found in eukaryotic cells.

· **Oligonucleotides -** A polymer made up of a few nucleotides.

· **Oncogene -** An oncogene is a gene that has the potential to cause cancer. In tumor cells, they are often mutated or expressed at high levels. Most normal cells undergo a programmed form of death.

· **Pathogenic -** Causing disease or abnormality.

· **Pathology -** Pathology is a significant component of the causal study of disease and a major field in modern medical practice and diagnosis.

· **Pedigree -** In medical genetics, a family history of a hereditary condition, or a diagram of a family history indicating the family members, their relationship to the proband, and their status with respect to a particular hereditary condition.

· **Penetrance -** Penetrance in genetics is the proportion of individuals carrying a particular variant of a gene that also expresses an associated trait.

· **Personalized medicine -** A medical model that proposes the customization of healthcare using molecular analysis - with medical decisions, practices, and/or products being tailored to the individual patient.

· **PET-scan -** A computerized radiographic technique that employs radioactive substances to examine the metabolic activity of various body structures.

· **Pharmacodynamics -** The effects of a drug or its metabolites on physiological function and metabolic pathways.

· **Pharmacogenomics -** Pharmacogenomics is the technology that analyses how genetic makeup affects an individual's response to drugs.

· **Pharmacokinetics -** The rate at which the body absorbs, transports, metabolizes, or excretes a drug or its metabolites.

· **Pharmacology -** The branch of medicine and biology concerned with the study of drug action, where a drug can be broadly defined as any man-made, natural, or endogenous (within the body) molecule which exerts a biochemical and/or physiological effect on the cell, tissue, organ, or organism.

· **Phenotype -** The observed biochemical, physiological, and morphological characteristics of an individual, as determined by his or her genotype and the environment in which it is expressed. Also, in a more limited sense, the abnormalities resulting from a particular mutant gene.

· **Point mutation -** A single nucleotide base pair change in DNA.

· **Polymorphism -** The occurrence together in a population of two or more alternative genotypes, each at a frequency greater than that which could be maintained by recurrent mutation alone. A locus is arbitrarily considered to be polymorphic if the rarer allele has a

frequency of 0.01 (1%), so that the heterozygote frequency is at least 0.02. Any allele rarer than this is a rare variant.

· **Polypharmacy -** The practice of administering many different medicines especially concurrently for the treatment of the same disease

· **Positive predictive value -** With respect to a clinical test for a disease, the extent to which testing positive indicates that one has or will develop the disease.

· **Prenatal -** Preceding birth.

· **Prevalence -** The number of cases of a specific disease present in a given population at a certain time.

· **Prodrugs -** A medication that is administered in an inactive or less than fully active form, and then it becomes converted to its active form through a normal metabolic process, such as hydrolysis of an ester form of the drug.

· **Prognosis -** Is a medical term for predicting the likely outcome of one's current standing.

· **Progression-free survival -** The length of time during and after the treatment of a disease, such as cancer, that a patient lives with the disease but it does not get worse.

· **Prophylactic mastectomy -** Prophylactic mastectomy is a surgery to remove one or both breasts to reduce the risk of developing breast cancer. According to the National Cancer Institute, prophylactic mastectomy in high-risk women may be able to reduce the risk of developing breast cancer by 90%.

· **Protein -** Proteins are large biological molecules, or macromolecules, consisting of one or more long chains of amino acid residues.

· **Proteomics -** A field of biochemistry encompassing the comprehensive analysis and cataloguing of the structure and function of all the proteins present in a given cell or tissue (see *proteome*).

Parallels *genomics,* a similarly comprehensive approach to the analysis of DNA sequence and mRNA expression.

· **Radiation therapy -** Treatment of disease with radiation, especially by selective irradiation with x-rays or other ionizing radiation and by ingestion of radioisotopes.

· **Resistance -** The reduction in effectiveness of a drug such as an antimicrobial or an antineoplastic in curing a disease or condition.

· **Ring chromosome -** A structurally abnormal chromosome in which the telomere of each chromosome arm has been deleted and the broken arms have reunited in ring formation.

· **RNA (ribonucleic acid) -** A nucleic acid formed on a DNA template, containing ribose instead of deoxyribose. *Ribosomal RNA (rRNA),* a component of the ribosomes, functions as a nonspecific site of polypeptide synthesis.

· **Sagittal plane -** A longitudinal plane which passes from anterior to posterior and divides the body into right and left sections.

· **Sanger sequencing -** Currently, the method most widely used to determine the nucleotide sequence of a DNA molecule. The DNA whose sequence is to be determined is used as a template for a polymerase that extends a complementary primer in the presence of four different dideoxynucleotides ("chain-terminating" nucleotides) corresponding to the four bases, ACGT, found in DNA. The length of the strands produced corresponds to which dideoxynucleotide was incorporated and terminated the extension reaction and therefore what base was present in the template at that site in the molecule.

· **Sequencing –**In genomics and molecular genetics, the order of nucleotides in a segment of DNA or RNA.

· **Serum marker -** A specific indicator found in a blood test that identifies a disease.

· **Sickle cell disease -** Sickle cell disease is a group of disorders that affects hemoglobin, the molecule in red blood cells that delivers oxygen to cells throughout the body. People with this disorder have

atypical hemoglobin molecules called hemoglobin S, which can distort red blood cells into a sickle, or crescent, shape.

· **Single gene disorder -** A hereditary disorder caused by a mutant allele of a single gene–eg, cystic fibrosis.

· **Single Nucleotide Polymorphisms (SNPS) -** A polymorphism in DNA sequence consisting of variation in a single base.

· **Spontaneous abortion -** A naturally occurring termination of a pregnancy.

· **SRY -** Symbol for a "maleness" gene found on the sex-determining region of the Y chromosome. The gene is believed to function as a master control switch that can turn off or on other genes involved in sexual development.

· **Strabismus -** A condition in which the eyes do not point in the same direction. It can also be referred to as a tropia or squint.

· **Stillbirth** - A stillbirth is defined as the death of a fetus at any time after the twentieth week of pregnancy.

· **Subtelomere -** Segments of DNA between telomeric caps and chromatin.

· **Systems biology -** The construction of analysis of complex data sets.

· **Systems medicine -** The application of biological concepts to improve patient care.

· **Tay-Sachs Disease –** It is a rare inherited disorder that progressively destroys nerve cells (neurons) in the brain and spinal cord. Tay-Sachsdisease is caused by mutation in the alpha subunit of the hexosaminidase A gene (HEXA).

· **T-cell -** T cells or T lymphocytes are a type of lymphocyte that play a central role in cell-mediated immunity.

· **Tetralogy of Fallot -** Tetralogy of Fallot is a condition that is characterized by several congenital heart defects occurring at once. They include:

- ventricular septal defect (Abnormal passageway between the right and left ventricles)
- displaced aorta
- narrowed pulmonary valve
- thickened right ventricle wall

· **Transcription factors -** One of a large class of proteins that regulate transcription by forming large complexes with other transcription factors and RNA polymerase; these complexes then bind to regulatory regions of genes either to promote or to inhibit transcription.

· **Transcriptomics -** The study of transcriptomes, the complete set of RNA transcripts produced by the genome at any one time.

· **Translocation -** The transfer of a segment of one chromosome to another chromosome. If two nonhomologous chromosomes exchange pieces, the translocation is *reciprocal.*

· **Triple Negative Breast Cancer –** A breast cancer testing negative for the expression of estrogen, progesterone, and HER2/neu growth receptors.

· **Triploidy -** The presence of three haploid sets of chromosomes, instead of two, in all cells; results in fetal or neonatal death.

· **Trisomy 13 -** A syndrome characterized by mental retardation and defects to the central nervous system and heart, caused by having three copies of chromosome 13.

· **Trisomy 18 -** a congenital condition caused by the presence of an extra chromosome 18, characterized by severe mental retardation and multiple deformities.

· **Trisomy 21 -** Down syndrome (DS), also known as trisomy 21, is a genetic disorder caused by the presence of all or part of a third copy ofchromosome 21.

· **Tumor -** An abnormal growth of tissues. Tumors can be cancerous (malignant) or noncancerous (benign).

· **Tumor suppressor gene -** A type of gene that makes a protein called a tumor suppressor protein that helps control cell growth. Mutations (changes in DNA) in tumor suppressor genes may lead to cancer.

· **Turner Syndrome -** Turner syndrome is a chromosomal disorder affecting females wherein one of the two X-chromosomes is defective or completely absent.

· **Ulysses Pact or Contract -** A freely made decision designed to bind oneself in the future.

· **Unbalanced Translocation -** Unbalanced translocation occurs when there is transfer of a fragment of one chromosome to another chromosome or when there is unequal exchange of chromosomal fragments between two nonhomologous chromosomes resulting in extra or missing genes

· **Uterine Fibroids -** Uterine fibroids are noncancerous (benign) tumors that develop in the uterus, a female reproductive organ.

· **XIST Gene -** X-Inactive Specific Transcript Gene.