**acrocentric:** Acrocentric chromosomes are those with the centromere very close to one end. The short arms (p) usually have small dot-like appendages on stalks, known as “chromosomal satellites”. In an acrocentric chromosome, the p arm contains repeating sequences, such as the nucleolar organizing regions. The acrocentric chromosomes in humans are 13, 14, 15, 21, and 22.

**aging:** Slides are subjected to dry heat and/or ethanol, in order to denature the proteins, to remove water and fixative from the preparations, and to enhance the hardness and adherence of the material to the glass. Chemical aging was developed as an alternative to dry-heat aging, in order to shorten the FISH protocol and preserve cell freshness (thus increasing hybridization efficiency) and chromosome architecture (thus allowing DAPI staining).

**autosome:** All chromosomes, excluding sex chromosomes are classified as autosomes. Humans have a total of 22 pairs of chromosomes.

**band:** The series of darkly and lightly stained stripes that appear across a chromosome when chemically treated and/or stained with certain dyes. Each chromosome has its own characteristic banding pattern that facilitates the specific identification of chromosomes or parts of chromosomes. The process used to achieve this result is called banding.

**banding:** Spread chromosomes are stained/banded, which allows their visualization and identification on a microscope. The most common banding method, which is referred to as G-band, is done with Giemsa stain.

**base pair:** A DNA nucleotide is comprised of a sugar, a phosphate group, and a nitrogenous base. There are four different nitrogenous bases in DNA which are adenine, cytosine, guanine, and thymine, which are abbreviated by as A, C, G, and T respectively. In base pairing, adenine always pairs with thymine, and guanine always pairs with cytosine. “Base pair” is abbreviated as “bp”.

**cell culture:** To achieve cell growth and division, leading to a good mitotic index for metaphase harvest. Cell culture methods vary with the tissue of origin, e.g. bone marrow, peripheral blood, solid tumors, and cell lines of various origins.

**cell division:** The mechanism by which cells multiply during the growth of tissues or organs. The type of cell division involved in the growth of the body is known as mitosis, and the type of cell division that produces sperm or ova in the testis or ovary is known as meiosis.

**centromere:** The constricted part of the chromosome separating the short arm (p) from the long arm (q). It is the attachment point for the mitotic spindle during cell division.

**chromosomal satellite:** (also called satellite II) It is a small mass of chromatin attached to the short arm of each chromatid of human acrocentric chromosomes by a relatively uncondensed stalk or secondary constriction. They do not always stain darkly by G-band
and may be difficult to see. Satellites on different chromosomes are often attracted to one another, causing the acrocentric chromosomes to be in satellite association. Not to be confused with satellite DNA (satellite I).

**Chromosome arm:** In metacentric, submetacentric and acrocentric chromosomes, chromosome body is divided into two halves by the centromere. The shorter part is called the short arm (p; petite) and the longer part is referred to as the long arm (q).

**Chromosome number:** In humans, the chromosome number is 23 pairs for a total of 46. Half of the chromosomes, 23 (22 autosomes and X) are inherited from the mother and half (22 autosomes and X or Y) from the father.

**Chromosome:** Means “dark body” in Latin. Thread-like structures composed of DNA containing all genetic information from both parents within the nucleus of most cells of the body. Different kinds of organisms have different chromosome numbers.

**Counterstain:** A second stain or dye that is of a contrasting color to a first specific stain or dye used to identify a particular target. Counterstains can be used to stain cells, nuclei or DNA, depending on the assay. For FISH, counterstains consisting of fluorophores are used to stain DNA a contrasting color to distinguish the background DNA from the specifically labeled probe hybridized to the target DNA. Commonly used counterstains for FISH include DAPI, Torpo3, and propidium iodide.

**Cytogenetics:** The study of the number, structure, behavior during cell division and effects of chromosomes and their abnormalities on the genotype and phenotype of the individual.

**DAPI:** 4,6 diamino-2-phenylindole, a DNA dye which fluoresces a blue color when exposed to ultraviolet light (UV) and is used to stain the nuclear or background DNA of the cell in FISH assays. DAPI is a good counterstain for FISH when using red, green and blue labeled chromosome painting probes.

**Deletion:** loss of a whole chromosome or part of a chromosome. The term usually refers to loss of part of a chromosome (e.g. a partial deletion). A terminal deletion refers to breakage and loss off the end of a chromosome; interstitial deletion is the loss of material from within the chromosome, and between its ends.

**Denaturation:** A crucial step prior to FISH. Paired up complimentary bases along the duplex nucleic acid strands are separated into single strands. The breakup of chemical bonds involved in complementary base pairing is usually produced by heat (although chemical means can be used). Slides and probes are denatured at a high temperature (usually 75-80°C or 167-176°F) before FISH.

**Diploidy:** A cell with a normal chromosome number including two sets of parental chromosomes (23 from each parent).

**DNA (deoxyribonucleic acid):** The long double-stranded chemical molecule found in the nucleus of cells and packaged into chromosomes. DNA contains the genetic code in the form of genes.

**Duplication:** A double copy of part of a chromosome resulting in an extra (abnormal) dose of the duplicated material.

**Euchromatin:** That part of the chromosomal DNA which is genetically active and pale staining with G- and C- banding. Euchromatin is relatively rich in GC base pairs. During interphase it is uncoiled and transcriptionally active whereas heterochromatin is condensed and inactive.
familial: Tendency to occur in more members of a family than expected by chance alone. For example, Down syndrome which has occurred more than once within a family because of an inherited chromosome translocation, can be said to be familial in that family.

FISH (Fluorescence in situ hybridization): A technique used for fine localization in genome mapping and in cytogenetics. The technique uses fluorescent molecules to detect the molecular hybridization of a DNA probe to a chromosome spread on a slide.

formamide: CH₃NO, FW = 45.04. The concentration of formamide is inversely proportional to the kinetics of the hybridization reaction, so that each increase of 1% in the formamide concentration lowers the Tm of a DNA duplex by 0.7°C. DNA hybridization in a solution containing 50-80% formamide lowers the effective temperature to 42°C compared to aqueous hybridization solution which requires a temperature of 68°C.

G-band: The common banding produced with Giemsa stain and used to routinely identify chromosomes.

gene rearrangement: A change in the gene structure or molecular arrangement within the chromosome. Hematopoietic somatic cells destined to become T-cells or B-cells, rearrange their T-cell receptor genes and immunoglobulin genes in a fashion that allows for the generation of receptor diversity. Gene rearrangement also refers to the rearrangement in genes that occurs as a result of chromosomal translocation or similar structural changes. For example, bcr gene rearrangement occurs as a result of a translocation between two chromosomes t(9;22)

gene: A sequence of DNA within a chromosome, containing the genetic information (genetic code) that is responsible for a particular bodily function or characteristic.

genome: Refers to the total genetic material in the chromosomes from a cell of an organism; it represents one haploid set of chromosomes with the genes they contain

haploid: The chromosome number (23) found in the parental gamete; half the diploid number.

heterochromatin: Chromosomal material which is condensed and genetically inactive during interphase. It consists of repetitive DNA sequences that are relatively rich in AT base pairs and is late replicating in the cell cycle. In metaphase chromosomes, it is dark staining with G- and C- banding.

hybridization: Base pairing of two single strands of DNA or RNA by hydrogen bonding between complimentary nucleotides.

ideogram: A schematic drawing of a chromosome