Interpretation of Karyotype (Practical based on Paper 3)

The Paris classification in 1971 and later in 1975 added greater accuracy to identification. This system is based on banding pattern obtained along the length of the chromosome. Each chromosome has a unique banding pattern and gives numbers to the bands of each chromosome.

Symbols for Chromosome Nomenclature:

The internationally accepted symbols to denote certain normal and abnormal chromosome features are as follows:

- 46XX---- Normal female
- 46XY---- Normal male
- A-G------ Chromosome groups
- 1-22------ Autosome numbers
- X, Y---- Sex chromosomes

Abbreviations

- del deletion
- dup duplication
- inv inversion
- p short arm
- q long arm
- r ring chromosome
- s satellite
- t translocation
- ter terminal
- + or - Placed before the chromosome number indicate addition (+) or loss (-) of a whole chromosome eg, +21 indicates an extra chromosome as in Down syndrome.
- Placed after the chromosome number + or – indicate increase or decrease in length of a chromosome part, eg, 5p- indicates loss of part of short arm of chromosome % as in *cri du chat* syndrome

Band Numbering

Bands can be defined as chromosome segments that can be clearly distinguished from the adjacent segments by appearing light or dark with one or more banding techniques.

Numbering of the bands was proposed during the Paris conference (1971). According to which:

A **landmark** is defined as any distinct morphological feature that can be used for identification of the chromosome. Telomeres, centromeres and a number of prominent bands are used as landmarks.

A centromere divides the chromosome into two arms, p (petite) for short arm and q (queue) for long arm.

A section of chromosome between the two landmarks is called **region.** These regions are numbered in both p and q arms. Region adjacent to centromere is designated as 1. Region numbers increase away from the centre.

The bands within the regions are numbered based on same rule.

Thus the position of a particular band can be designated by:

- Chromosome number
- Arm designation
- Region number
- Band number

For example,

First band in the second region of short arm of chromosome one can be written as 1p21.

High resolution banding reveals sub- bands. To indicate a sub band a ‘dot’ is used, followed by the number of sub- band (they are numbered sequentially from the centromere).

Eg: 14q32**.3 is a sub- band.**

When the sub-band is further sub-divided by still higher resolution banding, an additional digit is added, eg, 14q32.3.3

Some examples:

A. Abnormalities

1. Interpret the following formula:

- 46, XY, t (2;5) (q21; q31)

Answer:

Total number of chromosomes present = 46, male.

Reciprocal Translocation between chromosome 2 and 5. Breakage and reunion has occurred between long arm of 2nd chromosome, region 2, band 1 and long arm of 5th chromosome, region 3, band 1.

2. Write the abbreviation for duplication on chromosome number 1, long arm, between region 2, band 2 and region 5. Total number of chromosomes is 46, female.

Answer: 46, XX, dup (1) (q22qq25)

3. Turner's Syndrome

Answer: 45, X:

4. Interpret, 47, XXY:

Answer: Klinefelter's Syndrome

References

1. https://www.ncbi.nlm.nih.gov/Class/MLACourse/Original8Hour/.../chrombanding.html
2. TalwarPankaj, Manual of Cytogenetics in Reproductive Biology;Jaypee Brothers Medical Publishers, 2014,New Delhi.
3. https://ghr.nlm.nih.gov/primer/howgeneswork/genelocation