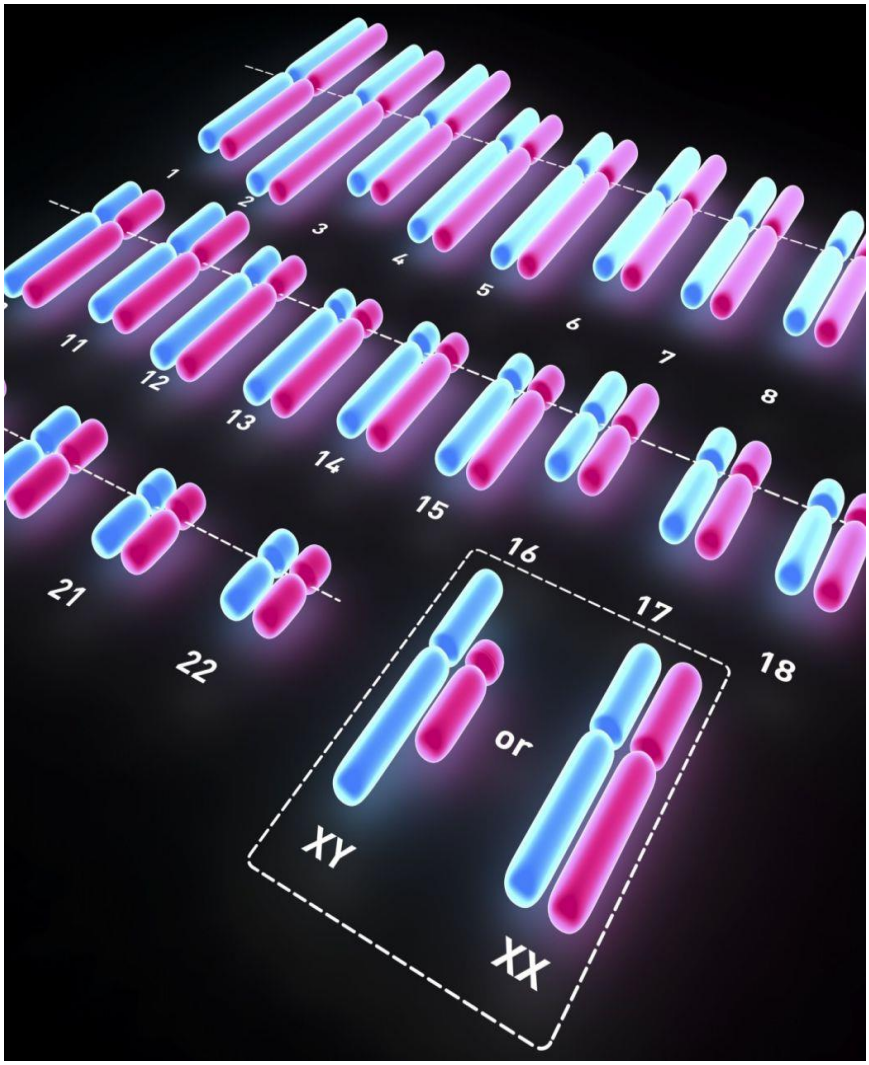

HUMAN GENETIC

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THIRD STAGE / EVENING STUDY

THEORETICAL PART

LECTURE NO. 4

21-11-2021

HUMAN GENETICS

3RD STAGE /MEDICAL LAB. TECHNIQUES
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The Karyotype and Chromosome Banding, Groups and Nomenclature

The Karyotype:

The karyotype may be defined as the phenotypic appearance of the chromosome complement and this includes both the number and morphology of the chromosomes, as revealed by the microcope at the metaphase stage of the cell division where the chromosomes are shortest and thickest The metaphase chromosome ,

Karyotypes of two species can differ in :

1. Chromosome number: Each species has a fixed diploid (2n)number of chromosomes . In Drosophila melanogaster , the diploid number is 8 chromosomes while in humans the 2n is 46 The diploid number(2n) is the number of chromosomes found in the somatic cells of the body The haploid number (n) of chromosomes is the one found in the gametes.
2. Chromosomes may vary in their length from long (> 10 um) to medium (4-8um) to short (> 2 um), and by the position of the centromere
3. The presence of a nucleolar organising region (NOR). This is a darkly stained region on the chromosome that represents the position of the nucleolus, NOR contains many tandemly repeated copies of the rRNA genes (185, 5.85 , 28S RNA genes) at this region.

Nucleolus is the place where ribosomes are built in eukaryotic. In humans , NORs occur in 5 homologous pairs.

4. Chromosomes may vary in the distribution of the heterochromatin Heterochromatin represents the highly condensed regions of the chromatin thread It is usually inactive and lacking genes as compared to the euchromatin which is less condensed and contains most of the active genes. Heterochromatin is usually found around the centromeres and sometimes at telomeres and called constitutive heterochromatin. It is, almost, always condensed and inactive

5. Chromosomes may vary in their staining patterns by the various cytological stains. Chromosomes may be stained by various stains to give various patterns of banding according to the stain used , and chromosome constituents

Chromosomes Banding:

Chromosomes (in mammals , including man) can be longitudinally differentiated by various staining techniques .

Staining the chromosome with the **malaria drug quinacrine** produces a series of bands that can be detected with an ultraviolet microscope where the stain will fluoresce at regions where the stain intercalates between the DNA bases Regions with less stain will not fluoresce intensively. These alternating fluorescent, and nonfluorescent regions are called the **Q-bands**.

If the chromosomes are pretreated with the enzyme **trypsin** that digests some of the chromosome proteins, and then stained with the, **Giemsa** stain a pattern of dark and light bands will be produced that can be visualized by the

ordinary microscope. These are the **G-bands** which correspond to the Q-bands , but G- bands can be detected by the light microscope while the Q-bands need an ultraviolet microscope;» Theretore , G-banding is the mostwidely used.

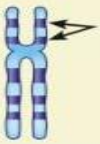
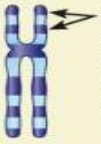
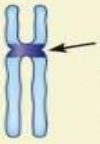
If we denature (by **heating**) the chromosomes and stain with **Giemsa** we will have another pattern of banding. In this pattern , the dark bands in the ordinary G - banding look light while the light ones look dark. In other words, this pattern is the reverse (R)of the G banding and hence it is called the **R-banding**.

If we denature the chromosomes with **barium hydroxide** and stain with **Giemsa** we get the centromere banding (**C- banding**) pattern. In this pattern the regions containing constitutive heterochromatin will stain dark These regions mostly found around the centromeres and sometimes at the telomeres The staining techniques and banding patterns can be summarized as in the following table

Table 1: The staining techniques and banding patterns.

Banding Technique	Procedure	Banding Patterns
G – banding	Mild proteolysis followed by staining with Giemsa	- Dark bands at AT- rich - Pale bands at GC- rich
R – banding	Heat denaturation followed by staining with Giemsa	- Pale bands at AT- rich - Dark bands at GC- rich
Q – banding	Stain with Quinacrine	- Dark bands at AT- rich - Pale bands at GC- rich
C – banding	Derature with barium hydroxide and then stain with Giemsa	- Dark bands contain constitutive heterochromatin

Table 2: The common chromosomes staining procedures.

Banding technique	Appearance of chromosomes
G-banding — Treat metaphase spreads with trypsin, an enzyme that digests part of chromosomal protein. Stain with Giemsa stain. Observe banding pattern with light microscope.	 Darkly stained G bands.
Q-banding — Treat metaphase spreads with the chemical quinacrine mustard. Observe fluorescent banding pattern with a special ultraviolet light microscope.	 Bright fluorescent bands upon exposure to ultraviolet light; same as darkly stained G bands.
R-banding — Heat metaphase spreads at high temperatures to achieve partial denaturation of DNA. Stain with Giemsa stain. Observe with light microscope.	 Darkly stained R bands correspond to light bands in G-banded chromosomes. Pattern is the reverse of G-banding.
C-banding — Chemically treat metaphase spreads to extract DNA from the arms but not the centromeric regions of chromosomes. Stain with Giemsa stain and observe with light microscope.	 Darkly stained C band centromeric region of the chromosome corresponds to region of constitutive heterochromatin.

Note:

1. These patterns could not be detected in other animals and plants. or, at least, they are not as clear as in mammals.
2. The most widely used patterns in karyotyping human chromosomes are the G-banding - and the R- banding
3. The convention in human cytogeneticsis to designate the long arm of the chromosome with letter (q) while the short arm is designated with letter (p). Numbering of regions or bands on each arm starts from the centromere toward the telomere of that arm
4. A human somatic cell (**diploid, 2n**) has 46 chromosome ; 44 of them are autosomes and 2 sex chromosomes. The sex chromosomes are identical in females and designated as XX while the male has one X and another different sex chromosome is Y (smaller than X)

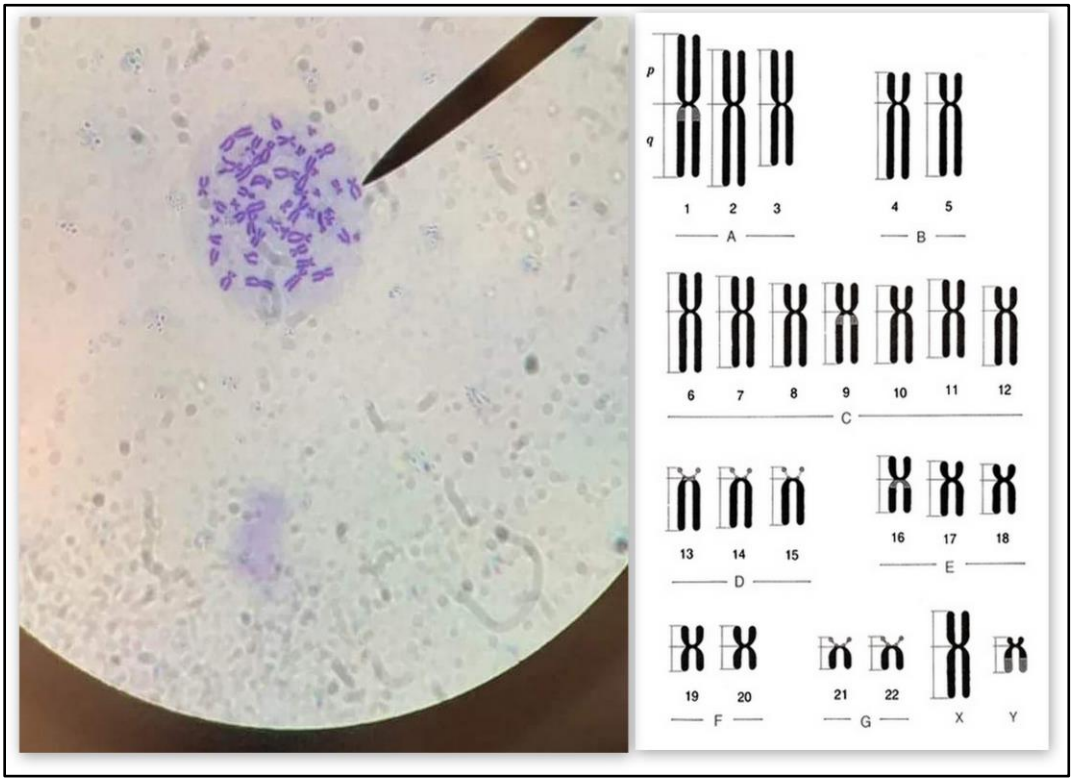
To write the karyotype:

1. Write the number of the chromosomes (46).
2. Write the sex chromosomes.

Therefore, the karyotype for a woman is **46XX** and the karyotype for a man is **46XX** Chromosomes occur in homologous pairs, for the autosomes (22 pairs) and a pair of (X) in the female (XX) and for the man the sex chromosomes are non-homologous (XY)

The haploid or the gamete (n) number is (22+X) for the woman and (22+Y) or (22+ X)for the man .

5. The 22 autosomal pairs are numbered in a decreasing order from (1) (the longest) to 22 (the shortest) . The sex (X , Y) chromosomes are not numbered.
6. To produce a chromosomal karyotype , stain (with a suitable stain) a cell at mitotic metaphase , photograph the chromosomes , cut, and arrange photographs.

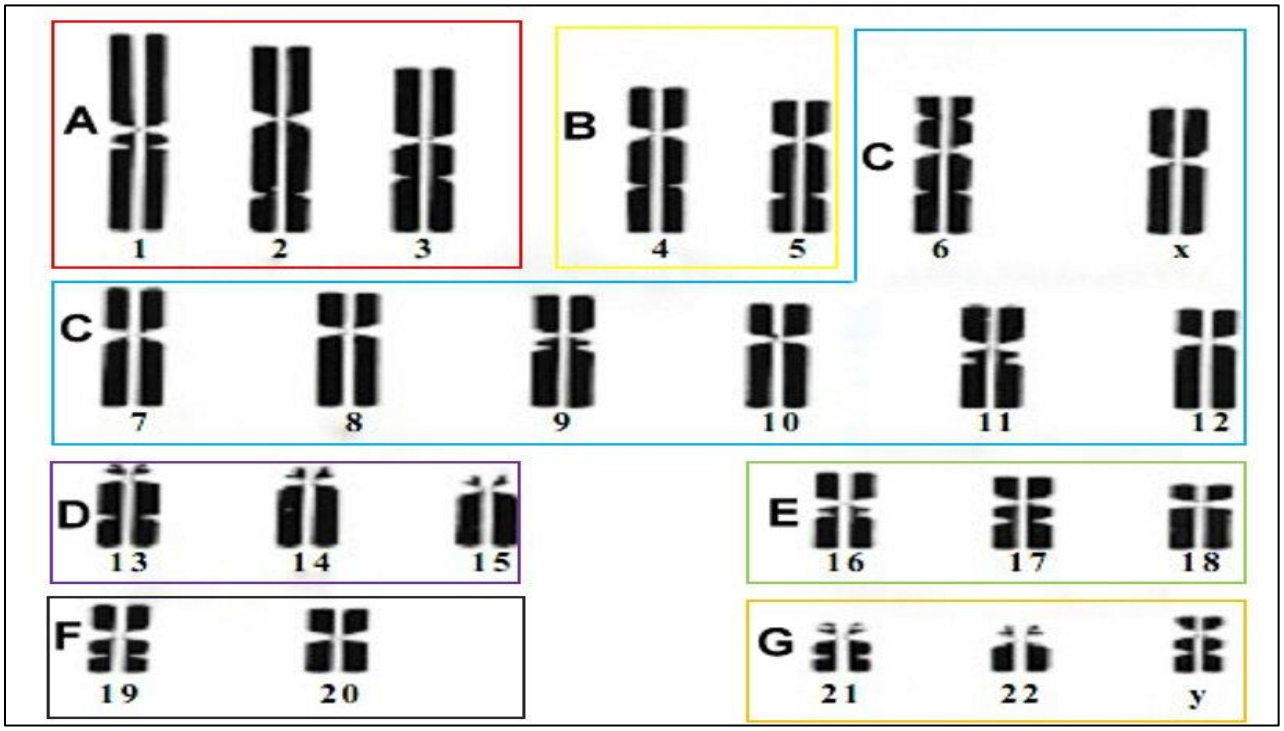


Chromosome groups

The 22 pairs are grouped (according to their length and the position of the centromes) into 7 groups (A, B, C, D,E, F and G). "A" contains the longest while "G' contains the shortest pairs. According to their sizes , X was put in the C group while the Y was put in the G group.

Table 3: The chromosomes groups.

Grouping	Number Of Chromosome	Description chromosome
Group A	Chromosome 1-3	Metacentric chromosomes are large and easily distinguished from the others because of its size and location of the centromere
Group B	Chromosome 4-5	has two large-sized chromosome sub metacentric
Group C	Chromosome 6-12, X	Metacentric chromosomes and medium-sized sub metacentric
Group D	Chromosome 13-15	acrocentric chromosomes of medium size and has satellite
Group E	Chromosome 16-18	Metacentric chromosomes and small-sized sub metacentric
Group F	Chromosome 19-20	Very small mmetacentric chromosome
Group G	Chromosome 21-22, Y	Acrocentric chromosomes are very small and have satellites except for Y chromosome



Chromosomes nomenclature

Human chromosome nomenclature is based on the results of several international conferences ISCN: An International System for Human Cytogenetic Nomenclature (1978)

Regions and bands are numbered consecutively from the centromere outward along each chromosome arm. Thus, the two regions adjacent to the centromere are labeled as 1 in each arm; the next, more distal regions as 2. and so on. A band used as a landmark is considered as belonging entirely to the region distal to the landmark and is accorded the band number of I in that region.

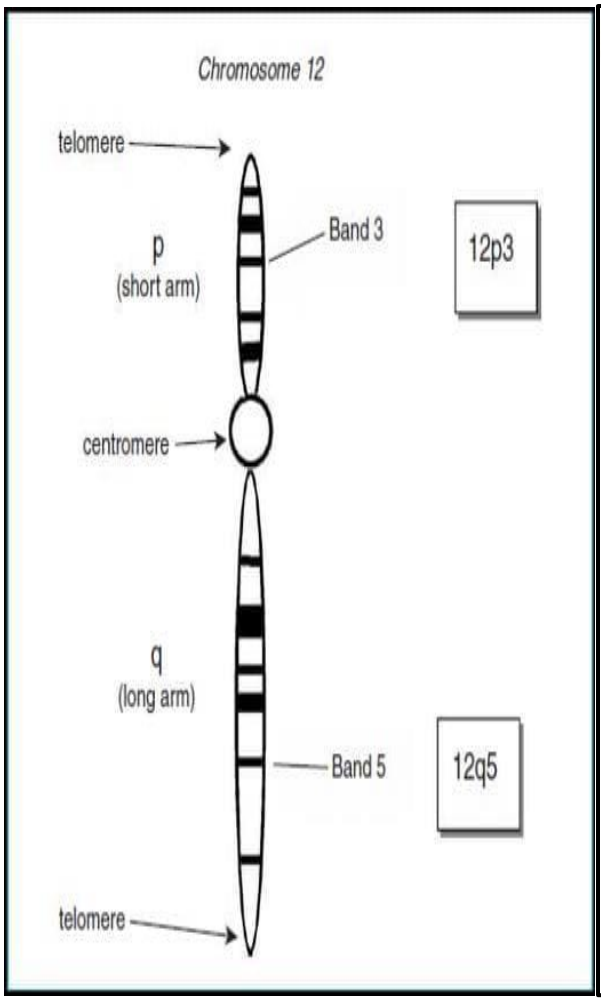
In designating a particular band, four items are required:

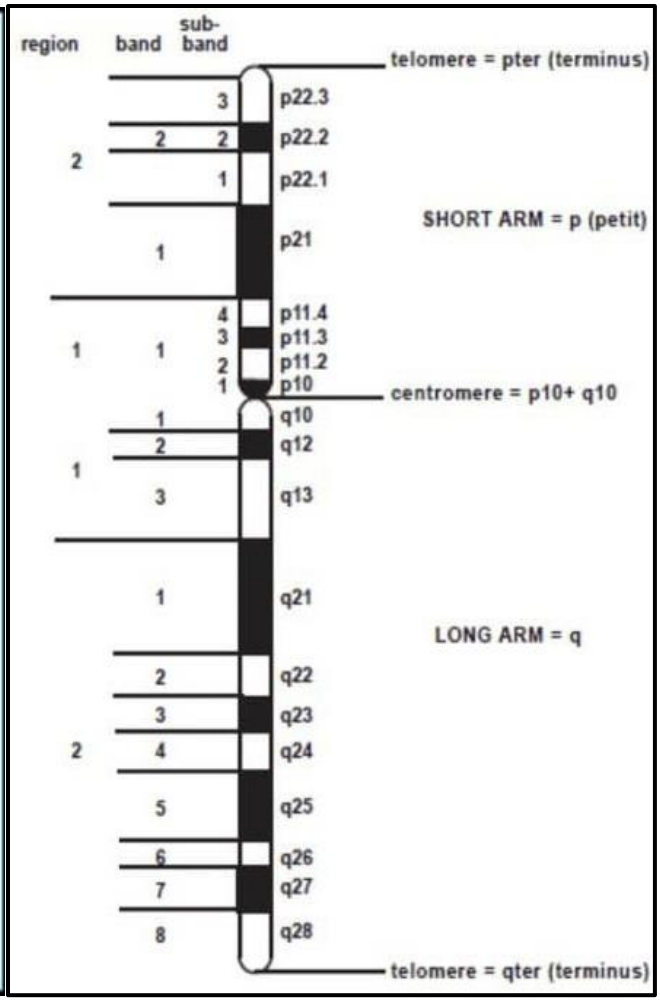
(1) the chromosome number.

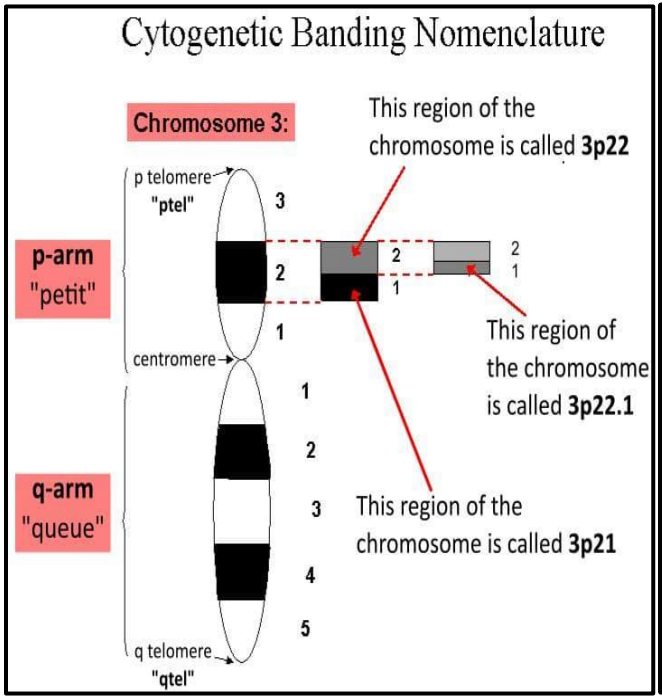
(2) the arm symbol,

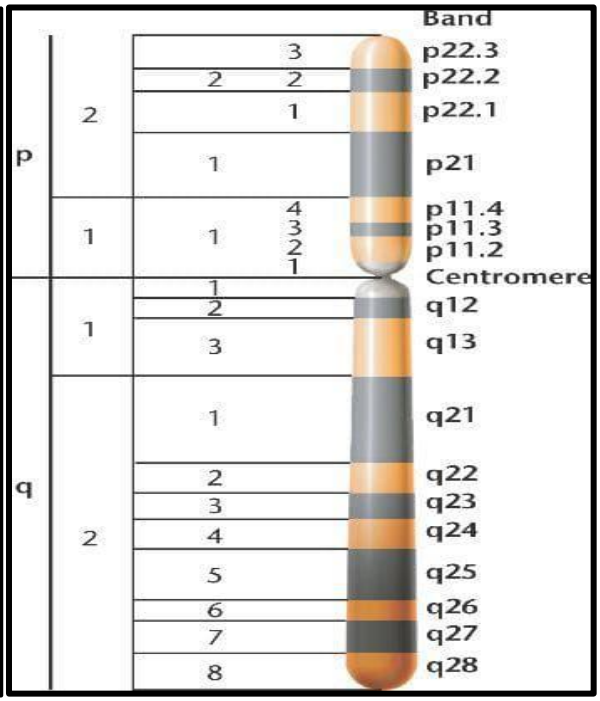
(3) the region number,

(4) the band number within that region. These items are given in order without spacing or punctuation. For example, Ip33 indicates chromosome 1, short arm, region 3, band 3.


Chromosome 12
telomere
p
(short arm)
Band 3
12p3
centromere
q
(long arm)
Band 5
12q5
telomere


region
band
sub-band
telomere = pter (terminus)
p22.3
p22.2
p22.1
SHORT ARM = p (petit)
p21
p11.4
p11.3
p11.2
p10
centromere = p10+ q10
q10
q12
q13
q21
LONG ARM = q
q22
q23
q24
q25
q26
q27
q28
telomere = qter (terminus)


Cytogenetic Banding Nomenclature
Chromosome 3:
This region of the chromosome is called 3p22
p telomere
"ptel"
p-arm
"petit"
centromere
This region of the chromosome is called 3p22.1
q-arm
"queue"
This region of the chromosome is called 3p21
q telomere
"qtel"


Band
p22.3
p22.2
p22.1
p21
p11.4
p11.3
p11.2
Centromere
q12
q13
q21
q22
q23
q24
q25
q26
q27
q28
p
q

Ex : name the following lable area?