Genetics

Biology Dept.3rd stage

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Lec-6-

KARYOTYPE:

Karyotype: is the general morphology of the somatic chromosome. Generally, karyotypes represent by arranging in the descending order of size keeping their centromeres in a straight line.

Idiotype: the karyotype of a species may be represented diagrammatically, showing all the morphological features of the chromosome; such a diagram is known as **Idiotype**

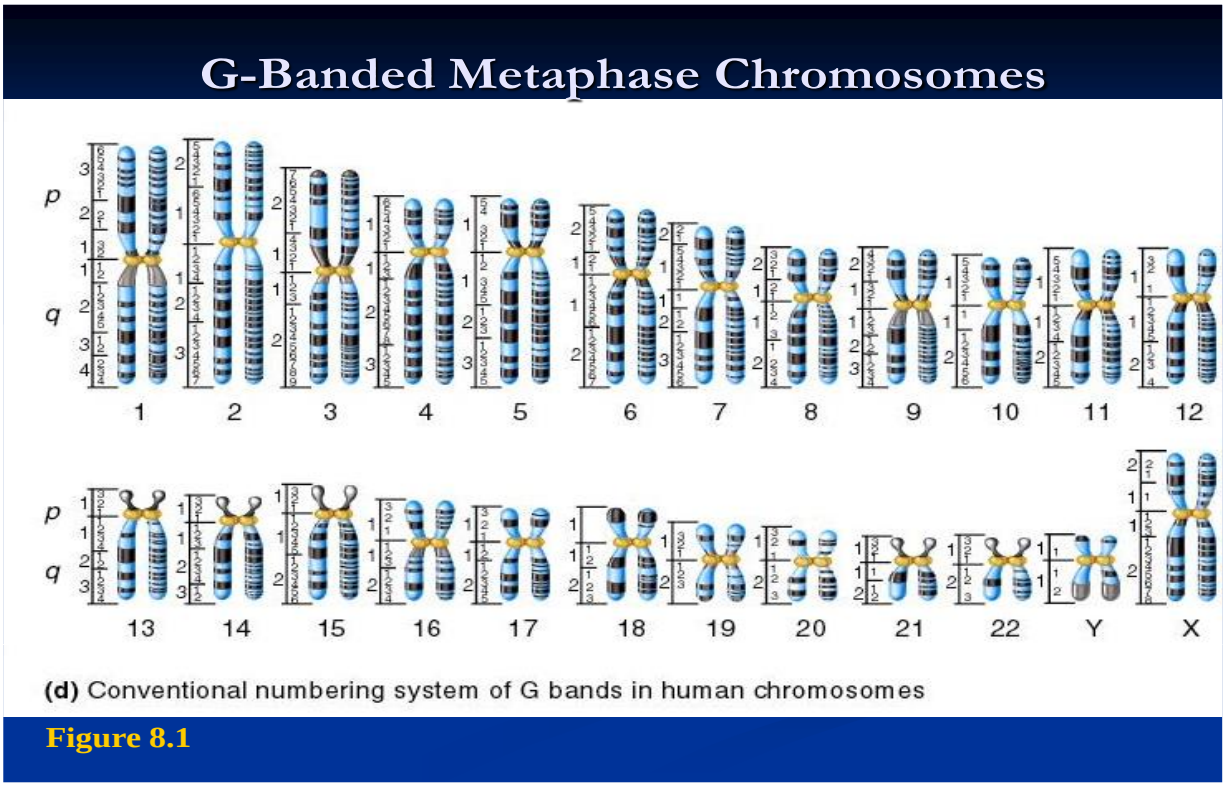

Figure 8.1

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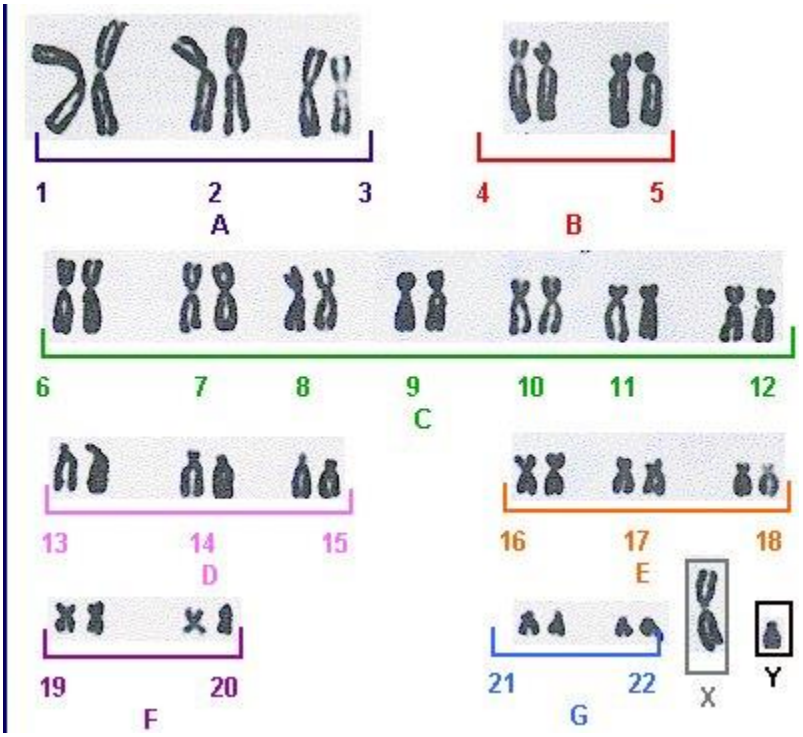

- 23 derived from each parent
- sex is determined by X and y chromosomes
- Males are XY
- Females are XX
- The sex of an offspring is determined by the sex chromosome carried in the sperm

Chromosomes are always arranged with the short arm on top

- Short arm is labeled P (French for petit)
- Long arm is labeled Q.

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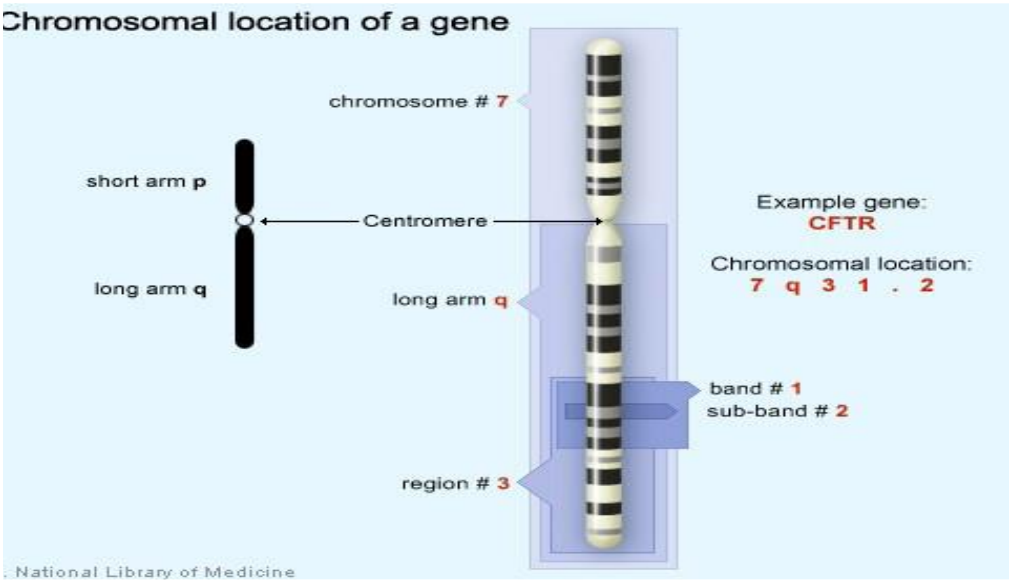


Chromosomes were first described by Strausberger in 1875.

The term "Chromosome", however was first used by Waldeyer in 1888.

They were given the name chromosome (Chromo = colour; Soma = body) due to their marked affinity for basic dyes.

Their number can be counted easily only during mitotic metaphase.

Chromosomes are composed of thin chromatin threads called Chromatin fibers.

These fibers undergo folding, coiling and supercoiling during prophase so that the chromosomes become progressively thicker and smaller during metaphase.

Normally, all the individuals of a **species have the same number** of chromosomes.

Closely related species usually have similar chromosome numbers.

Presence of a whole sets of chromosomes is called **euploidy**.

Gametes normally contain only one set of chromosome – this number is called **Haploid**

Somatic cells usually contain two sets of chromosome - **2n : Diploid**

The size of the chromosomes in mitotic phase of animal and plants sp generally varies between 0.5 µ and 32 µ in length, and between 0.2 µ and 3.0 µ in diameter.

In general, plants have longer chromosomes than animal and species having lower chromosome numbers have long chromosomes than those having higher chromosome numbers

Among plants, dicots in general, have a higher number of chromosome than monocots.

Euchromatin and Heterochromatin

Chromosomes may be identified by regions that stain in a particular manner when treated with various chemicals.

Several different chemical techniques are used to identify certain chromosomal regions by staining then so that they form chromosomal bands.

For example, darker bands are generally found near the centromeres or on the ends (telomeres) of the chromosome, while other regions do not stain as strongly.

The position of the dark-staining are heterochromatic region or heterochromatin.

Light staining are euchromatic region or euchromatin.

Euchromatin

Chromatin or chromosomal regions that are lightly staining, and relatively uncoiled during the anaphase . contain most of structural gene.

Heterochromatin

(a) Highly condensed region of chromosome
(b) Mainly located around centromere and telomere region
(c) About 10% of interphase chromosome
(d) Most part of mammalian Y chromosome is heterochromic
(d) Heterochromatin formation of one X chromosome in female cells

-Permanent inactivation for dosage compensation

(e) Nongenic regionHeterochromatin is classified into two groups: (i) Constitutive and (ii) Facultative.

Constitutive heterochromatin remains permanently in the heterochromatic stage, i.e., it does not revert to the euchromatic stage.

In contrast, **facultative heterochromatin** consists of euchromatin that takes on the staining and compactness characteristics of heterochromatin during some phase of development.

The structure of the interphase chromosome

(1) **Each interphase chromosome contains one DNA double helix**. (Unless it has passed through S-phase and then it has two double helices, joined at the **centromere region**. At this stage one can say that each chromatid has one DNA double helix.)

(2) A large proportion of the protein in chromatin consists of the proteins called **histones**. There are 5 major histone molecules.

(3) The **histone molecules** are basic (**positively charged**) proteins, which is why they associate so well with the negatively charged double helix.

(4) It is the **positively charged R-groups of lysine and arginine** that are most responsible for making histone positively charged. Please know the structure of the amino acid **lysine**:

(5) In the early 1970s, electron microscopists showed (with isolated and thinly "spread" chromatin) that the **primary structure of a eukaryotic chromosome appeared as "beads on a string**

(6) The beads were given the term **nucleosomes**.

(7) Chromosomes are composed of essentially equal masses of DNA and protein. This combination of DNA and protein was called chromatin by the early microscopists because it stained the same color (chromo) as the chromosomes with various stainsThe major proteins of chromatin are the histones – small proteins containing a high proportion of basic amino acids (arginine and lysine) that facilitate binding negatively charged DNA molecule proteins in eukaryotic cells.

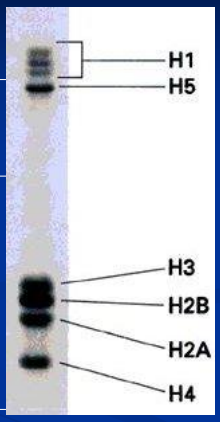
(8) Analysis of the nucleosome showed them to be composed of: (a) **2 "turns" of DNA double helix** around (b) **8 histone molecules.** The 8 histones are said to from an **octamer** (*oct* = 8, *mer* = parts).

(9) There were **2 molecules** each of the following **4 types of histone molecule**; **2A**, **2B**, **3**, and **4.**

Thus, both nuclease digestion and the electron microscopic studies suggest that chromatin is composed of repeating 200 base pair unit, which were called nucleosome.

The major histone proteins:

Histone	Mol. Wt	No. of Amino acid	Percentage Lys + Arg
H1	22,500	244	30.8
H2A	13,960	129	20.2
H2B	13,774	125	22.4
H3	15,273	135	22.9
H4	11,236	102	24.5

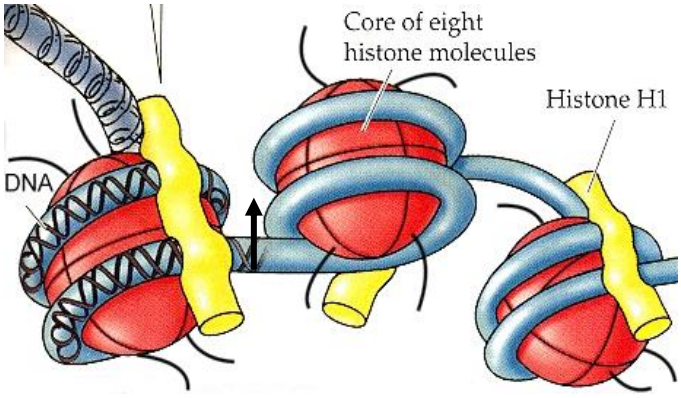


The DNA double helix is bound to proteins called histones. The histones have positively charged (basic) amino acids to bind the negatively charged (acidic) DNA. Here is an SDS gel of histone proteins, separated by size

Detailed analysis of these nucleosome core particles has shown that they contain 146 base pairs of DNA wrapped 1.75 times around a histone core consisting of two molecules each of H2A, H2B, H3, and H4 (the core histones).

(1) The nucleosomes are "pulled together" by the addition of another type of histone molecule (**histone 1**), to the outer surface of the nucleosome, and various non-histone proteins to the "linker" region of the DNA. The latter is not shown in the diagram below. The "beads on a string" chromatin "fiber" is about **10 nm in diameter**.

HISTONE 1



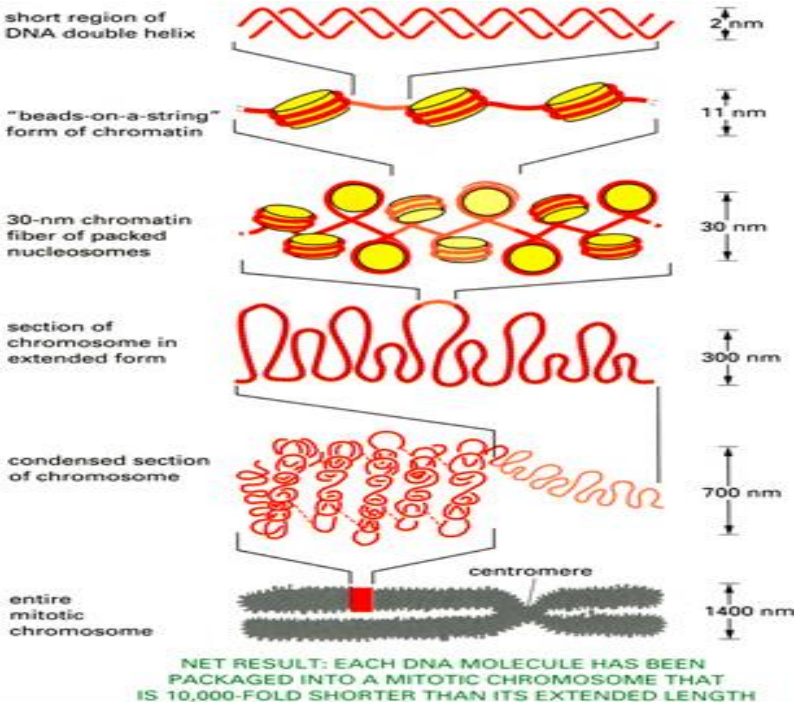
LINKER REGION

(2) This compacted "beads on a string" structure is now twisted to form a thicker "fiber" that is **30 nm in diameter**. Hence this fiber is about 3X thicker than the plasma membrane (which has a diameter of about 10 nm).

(3) This 30 nm diameter fiber is then folded (pleated) into loops when it is bound to various non-histone scaffold proteins.

(4) Depending upon the exact condensation state of the heterochromatin there can be further "packing" events.

(5) To form a mitotic chromosome another set of proteins called **condensins** is required.



There are 5 major types of histones: H1, H2A, H2B, H3, and H4 – which are very similar among different sp of eukaryotes. The histones are extremely abundant