Lecture 16 & 18

Chromosomal aberration: Variation in chromosome number – euploid, aneuploid, types of aneuploids and their origin; Nondisjunction - Klinefelter syndrome and Turner syndrome; Definition of eugenics and euthenics.

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NUMERICAL CHROMOSOMAL ABERRATIONS

- Each species of micro -organisms, plants and animals is characterized by particular chromosome complement or set of genome, represented once in gametic (haploid) cell i.e. n and twice in somatic (diploid) cells i.e. 2n.
- The term **genome refers to a complete set of chromosomes of a diploid species**.

- All the members of a genome are distinct from each other in gene content and often in morphology. Members of a genome do not pair.
- Possession of such sets of chromosomes or genomes, gives a specific chromosome number to each species.

- But sometimes, some irregularities may occur during mitosis, meiosis or fertilization and may produce cells with variant chromosome number.
- **A deviation from the diploid state represents a numerical chromosomal aberration which is often referred to as heteroploidy.**
- Individuals possessing variant chromosome number are known as **heteroploids**.

Numerical changes in chromosomes

- **A) Alterations in whole chromosome sets (Euploidy)**
- **B)Additions or subtractions of individual chromosomes (Aneuploidy)**
- **Euploidy** individuals having the chromosome number which is an exact multiples of the basic or genomic number
- **Euploidy** is the term for cells, tissues, and individuals with one complete chromosome set (monoploidy) or with whole multiples (diploidy, polyploidy) of the basic, monoploid number of chromosomes characteristic of the species.

Numerical changes in chromosomes

B) Additions or subtractions of individual chromosomes (Aneuploidy)

- **Aneuploidy** is the term for cells, tissues, and individuals with **excess or lacking one or few individual chromosomes**; It is a change in the number of chromosomes that can lead to a **chromosomal disorder**

– **hyperploidy** is a type of aneuploidy when there is an excess number of chromsomes (trisomics, tetrasomics)

– **hypoploidy** is another type of aneuploidy when one or more number of chromosomes are lacking.

Definition

- Euploidy **(Greek word; Eu = true or even; ploidy = unit)**
- The chromosome number is an exact multiple of the basic or genomic number

- Aneuploid
- Loss or gain of one or more particular chromosome occur within a set is called aneuploidy

A) Alterations in whole chromosome sets

Euploidy — **Multiples of basic chromosome set**

haploid

diploid

Aberrant euploidy: **more or less than normal number**

monoploid (1*n*)

triploid (3*n*)

tetraploid (4*n*)

pentaploid (5*n*)

hexaploid (6*n*)

} **polyploidy**

Euploidy types

- Monoploid - Organisms have one basic set of chromosome (X)
- Often weak and sterile
- Differ from haploids which carry half or gametic chromosome number (n)
- **Haploid:** Haploid is a general term used to designate the individuals or tissues with a gametic chromosome number i.e. n.

Euploidy types contd…

- Diploids- two copies of genome (2x)
- Normal diploids are known as disomics
- Regular bivalent pairing during meiosis
- Polyploids
- Any organisms with more than two sets of chromosomes or genomes

B) Additions or subtractions of individual chromosomes

–Aneuploidy

Aneuploidy

The loss or gain of one or more chromosomes from a genome

-**monosomy** the loss of one chromosome: 2n-1

-Double monosomy -Two non-homologous chromosomes missing 2n -1-1

-**trisomy** the addition of one chromosome: 2n+1

-Double trisomy-Two non-homologous chromosomes extra 2n+1+1

nullisomy the loss of one chromosome pair: 2n-2

-**disomy** addition of one chromosome in a haploid: n+1

-Tetra somy- one chromosome pair extra :2n +2

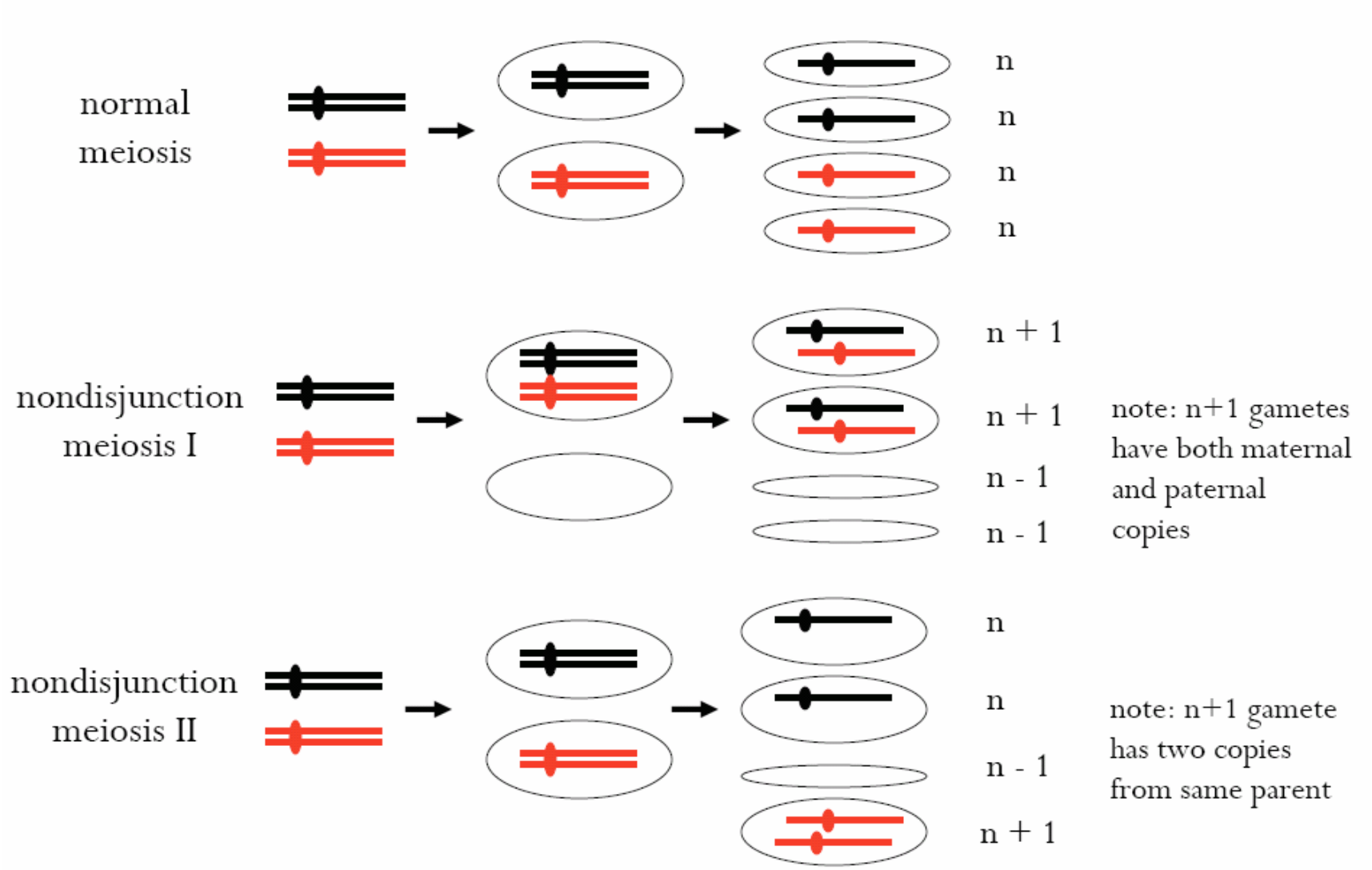
arise by nondisjunction during mitosis or meiosis-
failure of seperation of paired chromosomes

Origin of aneuploids

Non disjunction

- **Non-disjunction – failure of separation of paired chromosomes**
- occurs when paired chromosomes do not separate either during meiosis I or meiosis II.
- The direct result of this event is that gametes develop that have too few or too many chromosomes.
- If this occurs during meiosis I normal gametes are not developed, and if it occurs during meiosis II half of the gametes will be normal and the other half will be abnormal.

Non- disjunction of chromosomes



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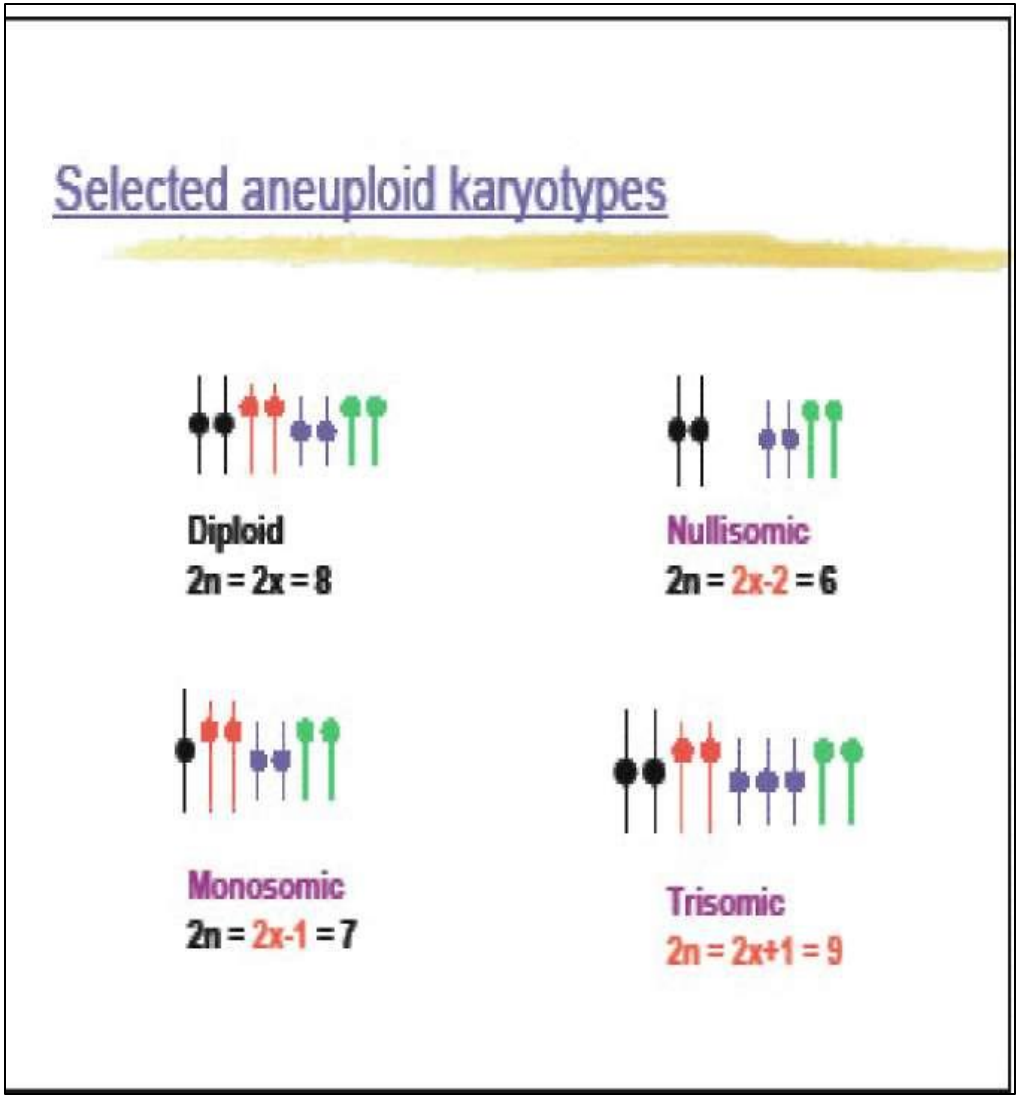


Types of Aneuploids

•**Trisomics -** presence of an extra chromosome of the standard set, (2n + 1)

•**monosomics -** loss of a single chromosome (2n -1)

•**nullisomics -** loss of a chromosome pair (2n - 2)

•**tetrasomics -** occurrence of an extra chromosome pair (2n + 2).

Sources of Aneuploids

1. Spontaneous occurrence from normal 2n plants, usually from random segregation of a univalent.
2. From asynaptic disomics and aneuploids – meiotic mutants.
3. Haploids and other polyploids – Unequal segregation from autopolyploids (2x X 4x).
4. Numerical nondisjunction from multivalent configurations may generate trisomics and monosomics – Interchange heterozygotes.

Asynapsis and Desynapsis

- **Asynapsis:** Failure of pairing of homologous chromososmes during zygotene stage of Prophase I of Meiosis I
- **Desynapsis:** Falling apart of paired chromosomes during diplotene stage of Prophase I of Meiosis I

Alien addition and substitution lines

- **Alien addition lines**

  A line of individuals or cells carrying additional chromosomes or parts of chromosomes

- **Alien Substitution lines**

  It refers to a line of plants in which one or more alien chromosomes from a certain donor species replace one or more chromosomes of a recipient species.

Uses of Aneuploids

- Used to determine the phenotypic effect of loss or gain of different chromosomes
- Used to locate genes on a particular linkage groups
- To find the homeology of chromosomes of different genomes
- Used to identify the chromosomes involved in translocation
- Used to produce alien addition and substitution lines especially to transfer/ replace a particular chrmosome for specific traits like resistance traits

Nullisomics (2n -2)

- **Nullisomics** (**2n-2)** Nullisomics are those missing one pair of chromosomes.
- Nullisomics are not viable in diploid and tetraploid species but are tolerated in some hexaploid species, such as wheat and oats.
- **Most of the nullisomics have reduced size and vigor throughout their life cycle.**
- In general, they have a smaller number of tillers and reduced plant height.

SOURCES OF NULLISOMICS

- The best sources of nullisomics are the progenies of monosomics and monotelosomics.
- A complete set of 21 nullisomic lines were isolated in wheat by E. R. Sears
- **uses of nullisomics**

  Nullisomics can be used to assign a gene to a specific chromosome

Aneuploids in human disease

- Vast majority lead to spontaneous abortions.

Monosomics (2n-1)

- **Turner's Syndrome** females have lost one of the X chromosomes (XO), sterile

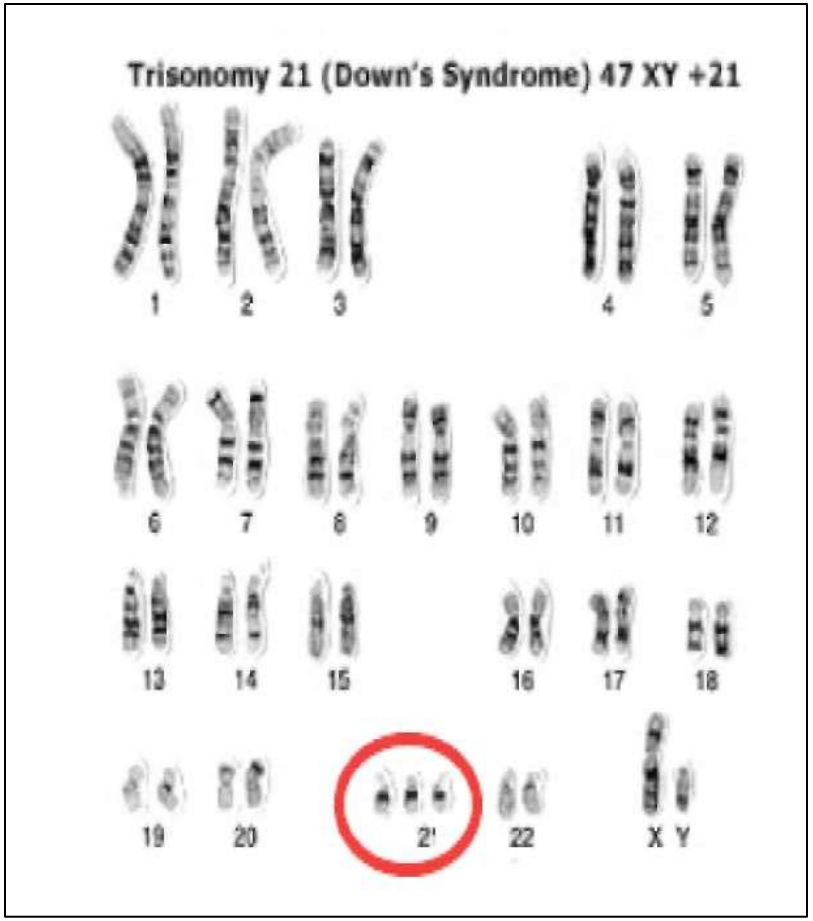
Trisomics (2n+1)

- **Klinefelter's Syndrome**: XXY feminized males; sterile

- **Down Syndrome:** Result of a trisomy of chromosome #21 (although a few cases due to a translocation)

- Trisomy 13 (Patau syndrome) and trisomy 18 (Edwards syndrome) can also survive to birth.

Trisomy(2n +1)

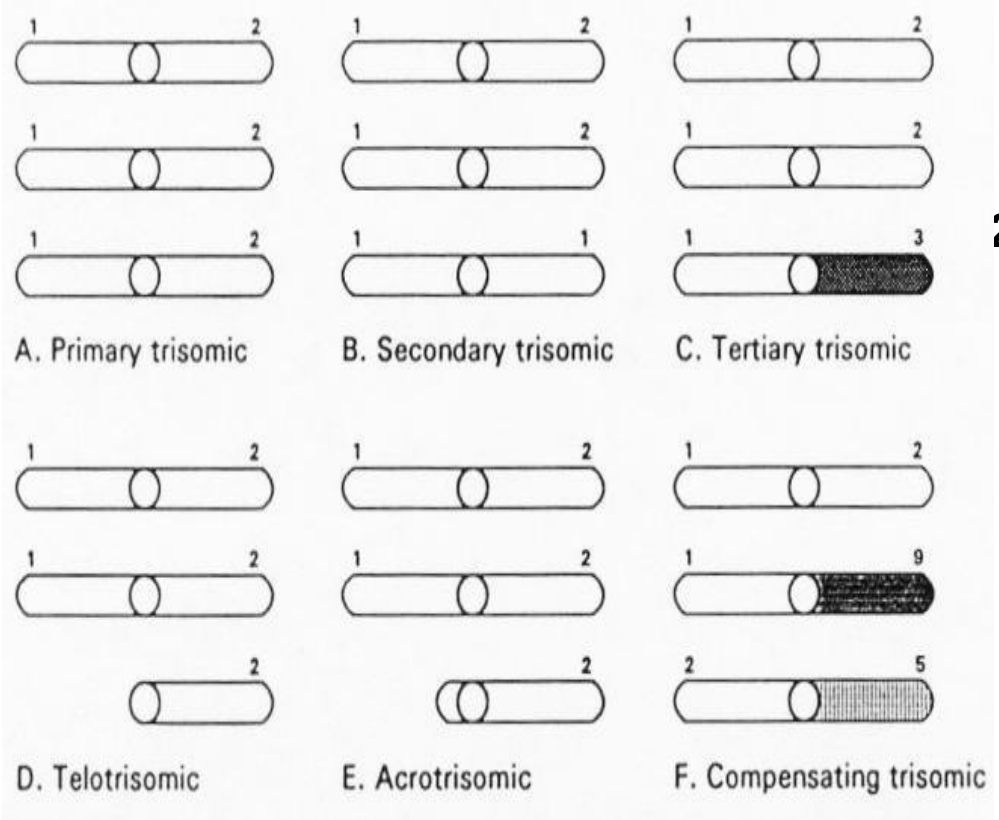


- A **trisomy** is the presence of three, instead of the normal two, chromosomes of a particular numbered type in an organism

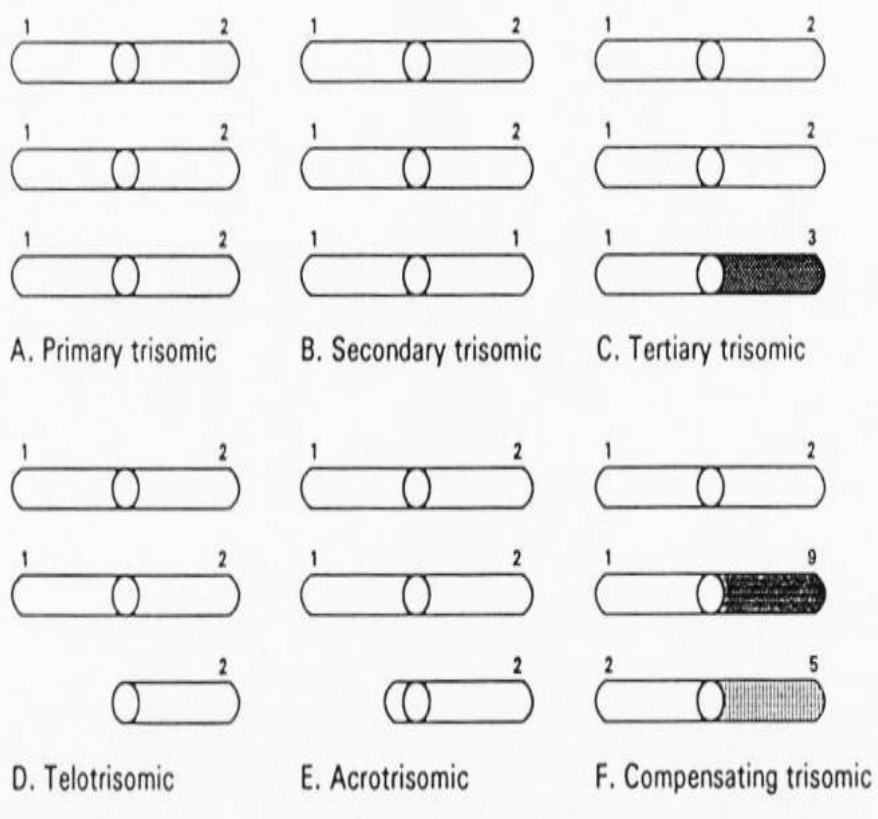
Origin of trisomics

- ***Secondary and telocentric trisomics***
- occurs occasionally among the progeny of normal plants
- but most frequently among progeny of plants that have one or more univalent chromosomes.
- They occur through the misdivision of the centromere.
- The best source is the progeny of primary trisomics.
- ***Tertiary trisomics*****:** among the progeny of translocation heterozygotes.

Types of trisomics



1. **Primary trisomics:** The additional chromosome is **completely homologous** to one of the chromosome pairs of the complement.
2. **Secondary trisomics:** The additional chromosome is a **isochromosome**.
3. **Tertiary trisomics**: The additional chromosome is a **translocated chromosome** derived from two nonhomologous chromosomes.



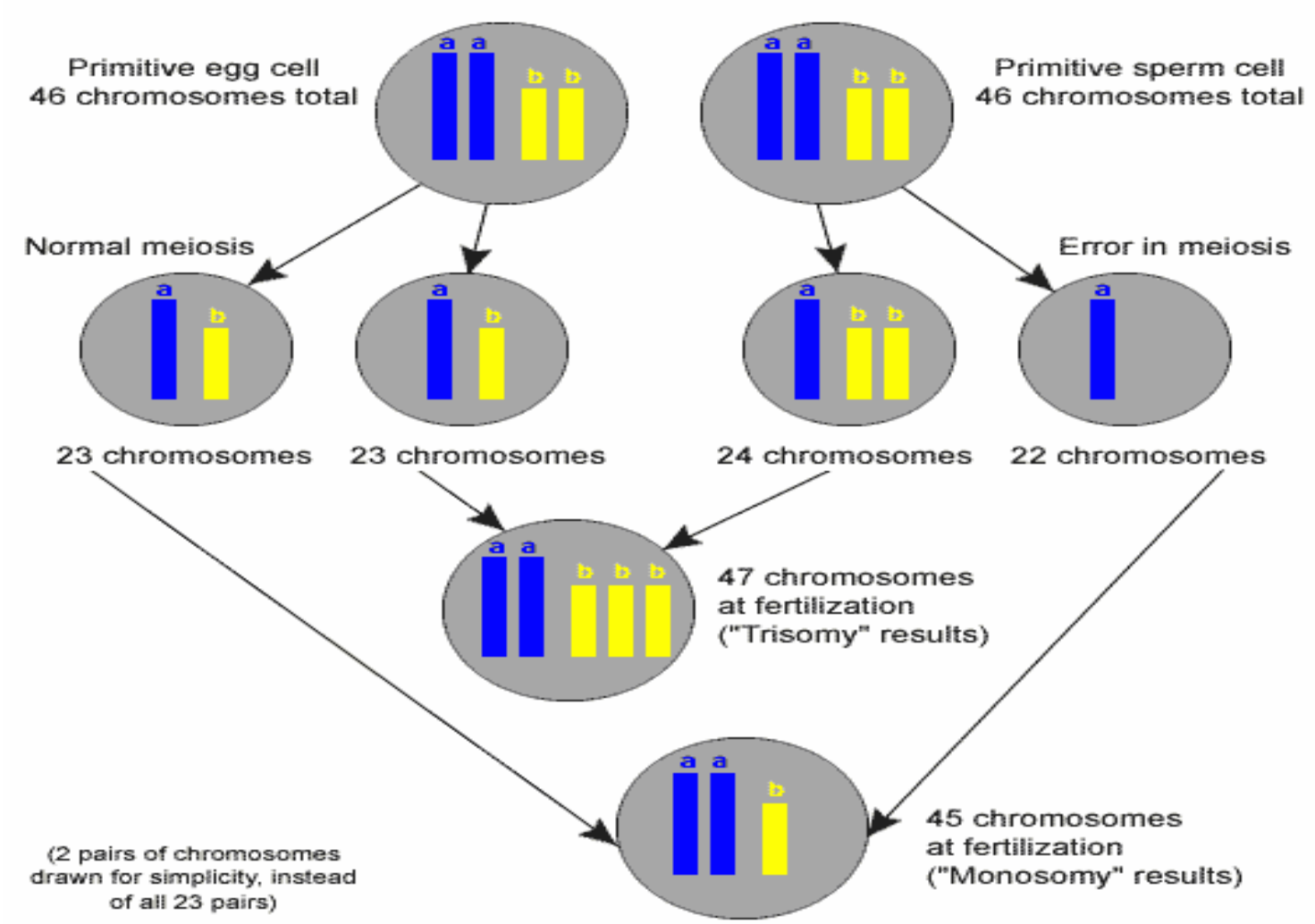
4. **Telocentric trisomics:** The additional chromosome is a telocentric chromosome.

5.**Acrotrisomic:** The additional chromosome is a acrocentric chromosome.

6.**Compensating trisomics:** A chromosome is missing and is genetically compensated by two other modified chromosomes.

Double trisomics: There are two different extra chromosomes (2n + 1 + 1).

Trisomoics in human beings



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Trisomy in human syndromes – Downs Syndrome

- 1 child in every 800-1,100 births has Down syndrome.
- 250,000 people in the U.S. have Down syndrome.
- Down syndrome results if the extra chromosome is number 21.
- The features of Down syndrome result from having an extra chromosome 21 in each of the body's cells.
- Down syndrome is also referred to as Trisomy 21, because of the presence of three number 21 chromosomes.

Klinefelter Syndrome – Trisomy

- The abbreviated name; XXY, KS
- Named after Dr. Henry Klinefelter (in 1942)
- Only a male inherited disease
- XXY feminized males; sterile
- It is a chromosomal disorder, is caused by nondisjunction disorder
- Born with an extra chromosome, 47 XXY
- Klinefelter's is a recessive disorder

Symptoms of Klinefelter's

- Taller than average height (longer limbs)
- 20-50% have mild intention tremors
- Smaller genitalia
- Poor upper body strength (clumsy)
- Poor Breast growth

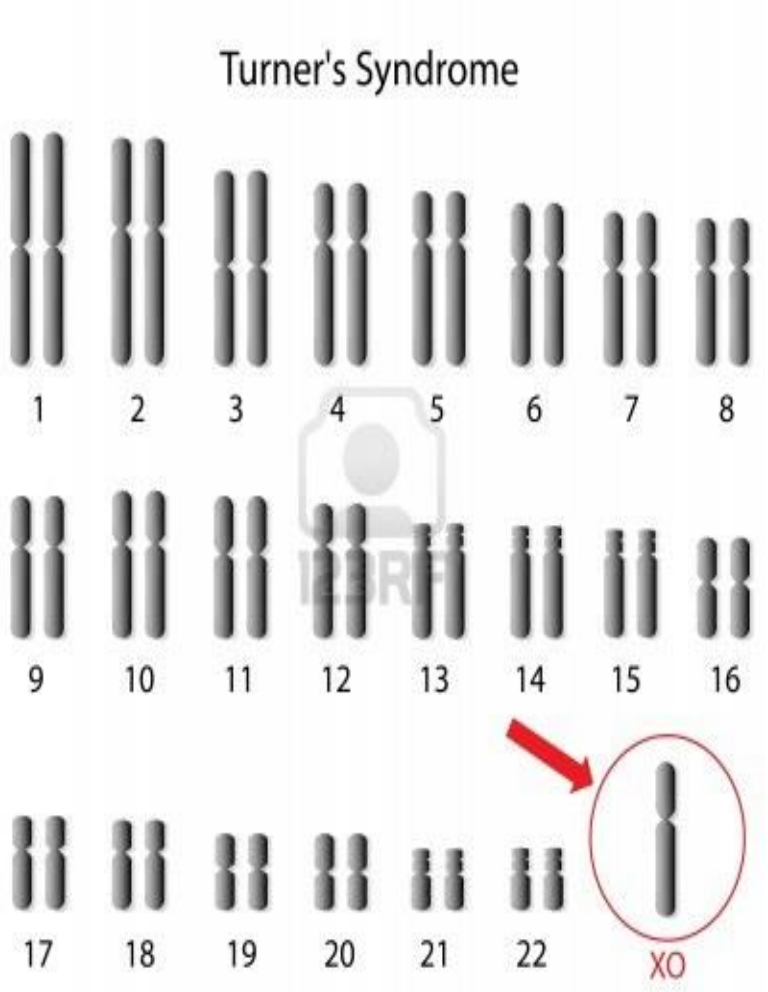
Applications of trisomics

- Analysis of dosage effects
- Assigning a new gene to a linkage group
- Linkage mapping: Primary trisomics are excellent tools for assigning linkage groups to specific chromosomes.
- The most extensive genetic studies with the aid of aneuploids have been conducted with trisomics.
- Assigning genes on specific arms using telocentric trisomics.
- Assigning molecular markers to specific chromosomes.

Monosomy (2n-1)

Presence of only one chromosome from a pair in a cell's nucleus

- The presence of only one
- individual with 2n - 1 chromosomes to produce n and n – 1 gametes in equal frequency



Origin of Monosomics

(1)Monosomics appear spontaneously in the progeny of normal disomics

(2)From asynaptic disomics and aneuploids. Some aneuploids, partially asynaptic due to the imbalanced chromosome number, constantly yield in their progenies monosomics for different chromosomes.

- For example, wheat nullisomic 3B is the major source for the isolation of various monosomics.

(3) From haploid plants

(4) From translocation heterozygotes

Sources of monosomics

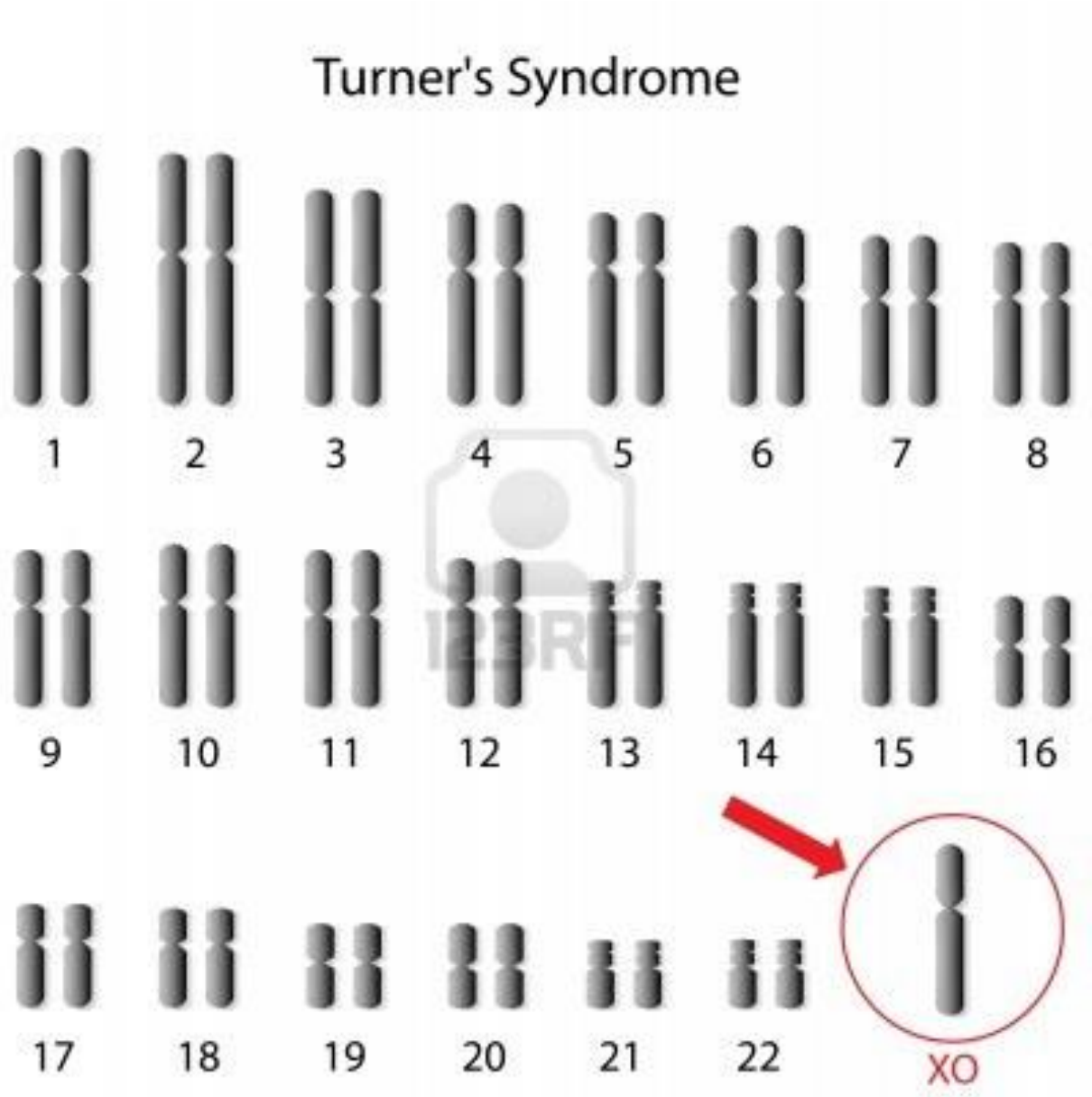
1. **asynapsis** as caused by nullisomy i.e. progeny of asynaptic nullisomic crossed to diploid.
2. **spontaneous haploid** progeny because meiosis in haploids is so irregular
3. chromosome loss as a result of **nondisjunction** during meiosis or early mitotic divisions of a diploid zygote
4. **unequal chromosome distribution -** non-co-orientation during meiosis of translocation heterozygotes
5. **treatment with various chemical (colchicine) or physical mutagens** which increases the incidence of above 1,3.

Applications of Monosomics

- ***Monosomic analysis: locating genes on specific*** chromosomes especially in polyploid spp *viz.,* cotton ,bread wheat etc. Locating recessive genes
- ***Identification of chromosomes belonging to the different genomes***
- ***Crosses of the various N. tabacum -*** monosomics with two parental species were used to identify the chromosomes of each genome.
- ***Mapping molecular markers on specific chromosomes***

Turner's Syndrome
Sterile females- Monosomy

- **Symptoms**
- Swollen hands/feet
- a wide/webbed neck (extra skin on neck)
- absence of puberty
- shield shaped chest; broad/flat
- drooping/ dry eyes
- infertility (due to absence of ovarian function)
- no menstruation cycle
- short in height


Turner's Syndrome
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Eugenics

- The term eugenics was coined in 1883 by **Francis Galton.**
- He coined the word eugenics to describe efforts at "race betterment." It comes from a **Greek word meaning "good in birth" or "noble in heredity."**
- In 1883, **Galton defined eugenics a**s "**The study of the agencies under social control that may improve or impair the racial qualities of future generations either physically or mentally.**

Types of eugeneics

- **Negative Eugenics:** preventing the births of children, with characteristics (genotypes/phenotypes) viewed as unhealthy or undesirable or preventing child bearing by "undesirable" individuals.
- **Positive Eugenics:** producing genetically enhanced children, giving them genetic characteristics (genotypes) they ordinarily would not be born with, and encouraging desirable individuals to bear more children.

Euthenics

- A science concerned with improving the well-being of mankind through improvement of the environment
- **Euthenics** : the study of methods of improving human well-being and efficient functioning by improving environmental conditions. (Or)
- Measures to improve the environment in order to improve health, appearance, behavior, or well-being of society.

Euphenics: Measures to improve the individual or phenotype (the body) by biological or medical means.