**Dr. S.K. Tiwari**

 **Zoology Department, DDU Gorakhpur University**

**Gorakhpur**

**--------------------------------------------------------------------------------------------------------------------**

 Numerical Chromosome Aberrations:

 **(for M.Sc. II Semester, Paper III)**

The gametic chromosome number constitutes a basic set of chromosomes called **genome**. A gamete cell contains single genome and is called **haploid**. When haploid gametes of both sexes (male and female) unite in the process of fertilization a **diploid** zygote with two genomes is formed. The diploid zygote undergoes embryological development and forms an adult animal which upon attaining sexual maturity produces haploid gametes. And this alternation of generation continues between haploidic and diploidic generation in most species. However, sometimes irregularities occur in nuclear division or “accidents” ( as from radiations) may befall inter- phase chromosomes so that cells or entire organisms with aberrant genomes may be formed. Such chromosomal aberra- tions may include whole genomes and entire single chromo- somes. Changes in number of whole chromosomes is called **heteroploidy** (see **Burns** and **Bottino**, 1989). Heteroploidy may involve entire sets of chromosomes **(euploidy**), or loss or addition of single whole chromosomes (**aneuploidy**). Each may produce phenotypic changes, modifications of pheno- typic ratio, or alteration of linkage groups. Many are of some evolutionary significance.

**GENETICS, HUMAN GENETICS AND EUGENICS**

**186**

**A. EUPLOIDY**

The term **euploidy** (Gr., *eu* = even or true; *ploid* = unit) designates genomes containing chromosomes that are multiples of some basic number (*x*). The euploids are those organisms which contain balanced set or sets of chromosomes in any number. The number of chromosomes in a basic set is called the **monoploid number**, *x.* Those euploid types whose number of sets is greater than two are called **polyploid**. Thus, 1*x* is **monoploid**, 2*x* is **diploid**; and the polyploid types are 3*x* (**triploid**), 4*x* (**tetraploid**), 5*x* (**pentaploid**), 6*x* **(hexaploid**) and so on. The **haploid** (**n**) refers strictly to the number of chromosomes in gametes. In most animals and many plants the haploid number and monoploid number are the same. Hence *n* or *x* (or 2*n* or 2*x*) can be used interchangeably. However, in case of polyploids the usage of *n* may create confusion. For example, in modern wheat *x and n* are different. Wheat has 42 chromosomes, but careful study reveals that in hexaploid there are six rather similar but not identical sets of seven chromosomes. So, 6*x* = 42, and *x* = 7. However, gametes of wheat contain 21 chromosomes, hence, 2n = 42 and n = 21 (see **Suzuki** *et al.,* 1986). A triploid hybrid of wheat, from which a hexaploid *Triticum spelta* has been obtained due to colchicine treatment, contains 2*n* = 3*x* = 21. Such haploids, since are obtained from the polyploids (*i.e.,* cross of tetraploid emmer wheat and diploid goat grass), they are called **polyhaploids**, just to differentiate them from the normal monoploids.

The lower organisms such as bacteria and viruses are called **haploids** because they have a single set of genetic elements. However, since they do not form gametes comparable to those of higher organisms, the term monoploid would seem to be more appropriate (see **Stansfield**, 1986).

1. **Monoploidy**

Certain insect species, such as ants and bees produce males that are derived from unfertil- ized eggs. These organisms are known as monoploids and are quite rare.

Monoploids have a single basic set of chromo- somes, *e.g.,* 7 in barley and 10 in corn. Monoploidy is common in plants and rare in animals.

* 1. **Origin and production of monoploids.** Mono- ploids in some cases are found normally and are produced due to parthenogenesis, as in male (drone) hymenopteran insects such as bees, wasps and ants. In these insects, queen and workers are diploid females. In angiosperms (flowering plants) monoploids may also originate spon- taneously due to parthenogenetic development of egg. Such rare monoploids have been obtained in tomatoes and cotton under cultivation. Rarely monoploid plants may originate from the pollen tube, synergids and antipo-

dals of the embryo sac and are called **androgenic mono- ploids** (or androgenic haploids).

Monoploids can be produced by artificial means by the following methods : (1) X-ray treatments, (2) delayed pollination, (3) temperature shock (cold treatment), (4)

colchicine treatment, (5) distant (interspecific or intergeneric) hybridization), (6) anther or pollen culture. Among these techniques, the most important ones are distant hybridization and anther culture.

1. **Distant hybridization.** Interspecific crosses in genera of *Solanaceae* (*e.g., Solanum* and *Nicotiana*) have been employed for the production of both parthenogenetic and androgenic mono- ploids. By this technique monoploids have been obtained in large number in potato. **Kasha** and **Kao** (1970) have used this technique for producing monoploids in large number in barley. They discovered that when diploid barley, *Hordeum vulgare*, is pollinated using a diploid wild relative called *Hordeum bulbosum***,** fertilization occurs, but during the ensuing somatic cell divisions, the chromosomes of *H. bulbosum* are preferentially eliminated from the zygote, resulting in a haploid embryo (such a **haplodization process** appears to be caused by a genetic incompatiability between the chromosomes

**CHROMOSOMAL MUTATION II**

**187**

of the different species). The resulting haploids can be doubled with colchicine treatment. This technique has resulted in the rapid production of new varieties of barley and applied to other plant species also.

1. **Anther or pollen culture.** The production of monoploids in tobacco plant by anther and pollen culture was demonstrated for the first time in the laboratory of **Prof. S.C. Maheshwari** of Dept. of Botany of Delhi University (**Guha** and **Maheshwari**, 1967). In this technique, a cell which is destined to become a pollen grain, may be induced by cold treatment to grow instead into an **embryoid**, a small dividing mass of cells. The embryoid may be grown on agar to form a monoploid plantlet, which can then be potted in soil to mature. Subsequently, such monoploids were produced for various crop plants such as soyabean, rice, wheat, mustard, and tobbaco. Presently, this technique is regarded as a very potential source of monoploid production.
	1. **Morphology of monoploids**. Monoploid plants have reduced size of all vegetative and floral parts. In monoploid *Nicotiana* **kostoff** reported that the leaves, flowers and over all plant size were smaller. The size of seed and stomata as well as diameter of pollen were found smaller in monoploids than in the diploids. Even the size of nucleus (or the nuclear volume) of a monoploid often was found to be just half than the nucleus of the diploid cell.
	2. **Cytology of monoploids.** In monoploids each chromosome is represented only once due to which there is no zygotene pairing and all the chromosomes appear as **univalents** on the metaphase plate at the time of meiosis. During anaphase each chromosome move independently of the other and goes to either of the two poles. According to law of probability the chance that a particular chromosome will go to a particular pole as half and the chance that all the chromosomes of a monoploid set will go to the same pole is ½ X ½ X ½. n times, where n = number of chromosomes in the monoploid set. So,

the frequency of gametes with the haploid set or n number of chromosomes will be (½)n. This indicates that higher the number of chromosomes in a haploid set, lesser will be the frequency of all of them being included in the same gamete. Gametes containing less than the haploid number of chromosomes are normally not viable. Therefore, monoploid organisms are highly sterile. For instance, a monoploid in maize (2n = 20) will have 10 chromosomes and the number of chromosomes in a gamete can range from 0–10. Consequently, considerable sterility is found in such monoploid maize plants.

In contrast, in monoploid male honey bees during spermatogenesis the meiosis is bypassed by mitosis. As a result, their sperms are haploid and viable.

* 1. **Uses of monoploids.** In a monoploid, since there is only one copy of each chromosome and only one allele of each gene, so, in it each gene is expressed whether it is dominant or recessive. This facilitates genetic experiments and this is the reason why microorganisms have been able helpful in genetic studies. For the same reason, scientists are trying hard to develop haploid strains of the flowering plants. Success has been achieved in developing monoploid strains of *Nicotiana*, *Datura* and *Triticum*. From these monoploid strains have also been developed pure breeding strains which are resistant for the insecticides and also for toxic compounds normally produced by the parasites of these plants.
1. **Polyploidy**

Any organism with more than two genomes (2*x*) is called a **polyploid**. Many plant genera include species whose chromosome numbers constitute a euploid series. For example, the rose genus *Rosa* includes species with the somatic numbers 14, 21, 28, 35, 42 and 56. These numbers are the multiples of 7. Therefore, this is a euploid series of the basic monoploid number 7, which gives diploid, triploid, tetraploid, pentaploid, hexaploid and octaploid species. Except diploids, rest of these belong to polyploid category. Ploidy levels higher than tetraploid are not commonly encountered in natural populations, but our most important crops and ornamental flowers are polyploids, *e.g.,* wheat (Hexaploid 6*x*), strawberries (octaploid, 8*x*), many commercial fruits and ornamental plants. Gener- ally, polyploidy is common in plants (more common in monocots) but rare in animals.

**GENETICS, HUMAN GENETICS AND EUGENICS**

**188**

**Types of polyploidy.** There are following three different kinds of polyploids : (i) autopolyploids,

1. allopolyploids and (iii) autoallopolyploids. Suppose that there are four different haploid sets of chromosomes A, B1 , B2 and C, in which B1 and B2 genomes are related. By using these genomes, all three types of polyploids can be derived as have been shown in Figure 15.1.



|  |
| --- |
| AAAA B1 B1 B1 B1auto-allooctoploidA B1 B1 B2hybirdAA B1 B1 CCAAAA A1AB1B1 B2 B2 B2 B2 allohexaploidautoetraploid allotetraploid B B B B autotetraploid1 1 2 2segmentaltetraploidAB1 Callotriploid hybridAA AB B1B1 B B B Bdiploid hybrid diploid 1 2 2 2 CChybrid diploid diploid**Fig. 15.1.** Mode of formation of different kinds of polyploid. |

* 1. **Autopolyploids.** The autopolyp- loids are those polyploids, which consist of same basic set of chromosomes multiplied. For example, if a diploid species has two similar sets of chromosomes or genomes (AA), an autotriploid will have three similar genomes (AAA), and an autotetraploid, will have four such genomes (AAAA).

Flower of one of the autoploid species of Chrysanthemum.

1. **Origin and production of auto- polyploids.** The autopolyploids may occur in nature or may be produced artificially. When they are found in nature, their auto- polyploidy nature is deduced by their mei- otic behaviour. One of the common ex- ample of natural autopolyploidy is ‘doob’ grass (*Cynodon dactylon*) which is quite commonly cultivated in U.P. and Bihar. Its **autotriploid** status was established from

its meiotic behaviour by **Prof. P. K. Gupta**, an eminent cytogeneticists of Northern India, working in Dept. of Agriculture Botany of Meerut University (**Gupta** and **Srivastava**, 1970). *Cynodon* is quite successful in the cultivation mainly due to its efficient way of vegetative propagation (since being triploid, it is sterile and setting no seed). Polyploids may arise naturally by following means : (i) in natural populations polyploidy may arise as a result of interference with cytokinesis, once chromosome replication has occurred; (ii) it may occur either in somatic tissues which give rise to tetraploid branches or during meiosis which produces unreduced gametes. All these natural inductions of polyploidy may occur due to chilling.

**CHROMOSOMAL MUTATION II**

**189**

|  |
| --- |
|  |
| Monoploid chromo- some set | Diploid (18) |
| Tetraploid (36)Hexaploid (54)Octaploid (72)Decaploid (90)Chromosome numbers in diploid and polyploid species of *Chrysanthemum.* Each set of homologous chromosomes is depicted in a different colour. |

Some of common examples of autotriploid crop plants, which are mainly produced by artificial methods, are seedless varieties of watermelons, sugar beet, tomato, grapes and banana. Similarly, many important crop plants include **autotetraploids** such as rye (*Secale cereale*), corn (*Zea mays*), red clover (*Trifo- lium pratense*), berseem (*Trifolium alexandrium*), marigolds (*Tagetes*), snapdragons (*Antirrhinum*), *Phlox*, grapes, apples, *Oenothera lamarkiana* (which was recognized as mutation by **Hugo de Vries)**.

**Induced autopolyploidy.** The autopolyploidy have been induced in many plant and animal cells by artificial means such as chemical (*e.g.,* chloral hydrate, colchicine, sulphanil amide, mercury chloride, hexachlorcyclohexane, etc.), radioactive sub- stances, *e.g.,* radium and X-ray) and temperature shocks. These inducers usually disturb the mitotic or meiotic spindle and cause non-segregation of already duplicated chromosomes, during cell divisions.

**Colchicine** is a drug (*i.e.,* an alkaloid obtained from the corms of plants–*Colchicum autmunale* and *C. luteum*) and its aqueous solution is found to prevent the formation and organiza- tion of spindle fibres, so the metaphase chromosomes of the affected cells (called **C-metaphase** or **colchicine metaphase**) do not move to a metaphase plate and remain scattered in the cytoplasm. Even the process of cytokinesis is prevented by colchicine and with duplications of chromosomes the number goes on increasing. As colchicine interferes with spindle forma- tion, its effects are limited to dividing and meristematic cells.

1. **Effects of autopolyploidy.** Autopolyploidy results in **gigantism** of plant cells, *i.e.,* leaves, flowers and fruits of an autopolyploid are larger in size than a diploid plant. For example, the size of lower epidermis of leaf of a tetraploid *Saxifraga pensylvanica* was found greater than the diploids (Fig. 15.2). Some of significant effects of autopolyploidy are as follows : (1) With the increase in cell size, the water content increases which leads to a decrease in osmotic pressure. This results into loss of resistance against frost, etc. (2) Due to slower rate of cell division, the plant’s growth rate decreases. This leads to a decrease in **auxin** supply and a decrease in respiration. 3. Due to slow growth rate, the time of blooming of an autopolyploid is delayed. 4. At higher ploidy level, such as autooctoploids, the adverse effects become highly pronounced and lead to the death of the plants.

Polyploid varieties with an even number of genomes (*e.g.,* tetraploids) are often fully fertile (Fig. 15.3), whereas those with an odd number (*e.g.,* triploids) are highly sterile (Fig. 15.4).

**Uses of induced polyploidy.** Since in the induced polyploids, the fertility level and seed set are low, so seedless fruits can be produced by using triploids as in case of seedless watermelons which were produced by a Japanese scientist, **Dr. Hitoshi Kihara**. These triploids are obtained from seeds raised by a cross of tetraploid and diploid plants. The tetraploids have been produced from the diploids by colchicine treatment. By adopting these methods a variety of triploids such as sugar beet, tomato

**GENETICS, HUMAN GENETICS AND EUGENICS**

**190**

and grapes and tetraploids such as rye, barley, corn, apple, grapes, marigolds, snapdragons, lily, phlox, etc., have been obtained. Among the forage crops, tetraploid barseem is a very popular crop in Northern India.

* 1. **Allopolyploids.** When the polyploidy results due the doubling of chromosome number in a F1 hybrid which is derived from two distinctly different species, then, it is called **allopolyploidy** and the resultant species is called an allopolyploid. Let A represent a set of chromosomes (genome) in species X, and let B represent another genome in a species Y. The F1 hybrids of these species then would have one A genome and another B genome. The doubling of chromosomes in the F1 hybrids will give rise to allotetraploids with two A and two B genomes (see Fig. 15.5).

*Raphanobrassica* is a classical example of allopolyploidy or amphipolyploidy. In 1927, a Russian geneticist, **G.D. Karpechenko** performed a cross between radish (*Raphanus sativum,* 2n = 18) and cabbage (*Brassica oleracea,* 2n = 18) and in F1 got sterile (diploid) hybrids. Among these sterile



|  |
| --- |
| epidermal cellsguard cells of stomata**A B****Fig. 15.2.** Comparison of size of leaf epidermal cells of a diploid (A) and tetraploid (B) saxifrage (after Burns and Bottino, 1989). |

F1 hybrids, he found certain fertile plants which were found to contain 36 chromosomes. These fertile tetraploids were called *Raphanobrassica.*

**Synthesized Allopolyploids**

To find out the origin of naturally occurring allopolyploids some cytogeneticists produced certain allopolyploids in laboratory by employing artificial means. Common hexaploid wheat and tetraploid cotton furnish two such examples.

**CHROMOSOMAL MUTATION II**

**191**

|  |
| --- |
| two bivalents11. pairing one
2. possibilities quadrivalent 4

1* 1. univalent

+* 1. trivalent

4**Fig. 15.3.** Meiotic pairing possibilities in tetraploids (each chromosome is really two chromatids). |
| trivalent11. pairing

possibilities1. bivalent

+univalent**Fig. 15.4.** Meiotic pairing possibilities in triploids (each chromosome is really two chromatids). |

|  |
| --- |
| **P1 :** Species X X Species Y(AA) (BB)(Diploid)  (Diploid)**F1 :** ABDiploid sterile hybridColchicineAA BBAmphidiploid tetraploid (Fertile)**Fig. 15.5.** Formation of an amphidiploid tetraploid. |

**GENETICS, HUMAN GENETICS AND EUGENICS**

**192**

1. ***Triticum spelta*** is a hexaploid wheat which was artificially synthesized in 1946 by **E.S. McFadden** and **E. R. Sears** and also by **H. Kihara**. They crossed an emmer wheat, *Triticum dicoccoides,* (tetraploid : 2n = 28) with goat grass, *Aegilops squarrosa* (diploid ; 2n = 14) and doubled the chromosome number in the F1 hybrid (Fig. 15.6). This artificially synthesized hexaploid wheat was

|  |
| --- |
| **P1 :** *Triticum dicoccoides* X *Aegilops squarrosa*(Tetraploid emmer wheat) (Diploid goat grass) AA BB DD(2n = 28 ; 14 bivalents)  (2n = 14 ; 7 bivalents)**F1 :** ABDTriploid hybrid(2n = 21 ; 21 univalents)Colchicine AA BB DDSynthesized hexaploid wheat(*Triticum spelta*) (2n = 42 ; 21 bivalents).**Fig. 15.6.** Artificial synthesis of hexaploid wheat. |

found to be similar to the primitive wheat *T. spelta.* When the synthesized hexaploid wheat was crossed with naturally occurring

The pods of the amphidiploid form of *Gossypium,* the cultivated cotton plant.

*T. spelta,* the F1 hybrid was completely fertile. This suggested that hexaploid wheat must have originated in the past due to natural hybridization between tetraploid wheat and goat grass followed by

subsequent chromosome doubling.

1. ***Gossypium hirsutum,*** the New world cotton plant, is another interesting example of allopolyploidy. Old world cotton, *Gossypium herbaceum,* has 13 pairs of chromosomes, while, Ameri- can or “upland cotton” also contains 13 pairs of chromosomes. **J.O. Beasley** crossed the old world and American cottons and doubled the chromosome number in the F hybrids. The allopolyploids, thus,

1

produced resembled the cultivated New world cotton and when crossed with it gave fertile F1 hybrids (Fig. 15.7). These results, thus, suggested that tetraploid *Gossypium hirsutum* originated from two diploid species, namely *G. herbaceum* (2n = 26) and *G. raimondii* (2n = 26).

|  |
| --- |
| *Gossypium herbaceum* X *Gossypium raimondii*(Old world cotton) (American or upland cotton) (2n = 26 ; 13 bivalents) (2n = 26 ; 13 bivalents)F1 hybrid(2n = 26; 26 univalents)ColchicineNew world cotton (*Gossypium hirsutum*) (2n = 52 ; 26 bivalents)**Fig. 15.7.** Artificial synthesis of new world cotton. |

**CHROMOSOMAL MUTATION II**

**193**

1. **Triticale *(Triticosecale Wittmack)*** is the first man made cereal which has been developed in recent years and is cultivated on about one million hectares of land throughout the Globe for the commercial use. Triticale is an artificial allopolyploid which has been derived by crossing wheat (*Triticum*) and rye (*Secale*). Depending upon whether *Triticum* is a tetraploid (2n = 4*x*= 28) or hexaploid (2n = 6*x* = 42), one would get hexaploid triticale (2n = 6*x* = 42; Fig. 15.8) or octaploid triticale (2n = 8*x* = 56; Fig. 15.9), respectively. In each case, only diploid rye (2n = 4*x* = 14) was used.

|  |
| --- |
| *Triticum durum* X *Secale cereale*(Tetraploid wheat ; 2n = 28) (Diploid rye ; 2n = 14)F1 hybrid (sterile) (Triploid ; 2n = 21)(Chromosome doubling)Hexaploid triticale (2n = 42)**Fig. 15.8.** Artificial synthesis of a hexaploid triticale. |

* 1. **Segmental allopolyploids.** Different genomes of some allopolyploids are not quite different from each other. Consequently in these polyploids chromosomes belonging to different genomes do pair together to some extent. This indicates that segments of chromosomes and not the whole chromosomes are homologous. Therefore, such allopolyploids are called **segmental allopolyploids (Stebbins**, 1943, 1950). The segmental allopolyploids are intermediate between autopolyploids and allopolyploids and can be identified by their peculiar meiotic behaviour.

It is generally believed that most naturally occur- ring polyploids are segmental allopolyploids. Our com- mon hexaploid bread wheat too is found to be a segmen- tal hexaploid.

**Polyploidy in animals.** Polyploidy is rare in animals but occur in flatworms, leeches and brine shrimp. In mice, also, 40 per cent liver cells are tetraploids, and about 5 per cent are octoploids. Polyp- loidy in humans have been found in liver cells and cancer cells. In them polyploidy is whether complete or as a mosaic, it leads to gross abnormalities and death.

*Triticum aestivum.*

Octoploid triticale (2n = 56)

**Fig. 15.9.** Artificial synthesis of a octoploid triticale.

doubling)





F1 hybrid (sterile) (Tetraploid ; 2n = 28)

(Chromosome

*Secale cereale*

(Diploid rye; 2n = 14)

X

*Triticum aestivum*

(Hexploid wheat; 2n = 26)

**GENETICS, HUMAN GENETICS AND EUGENICS**

**194**

**Phenotypic Effects of Polyploidy**

The increase in the genome’s size beyond the diploid level is often caused following detectable phenotypic characteristics in a polyploid organism :

1. **Morphological effect of polyploidy.** The polyploidy is invariably related with **gigantism**. The polyploid plants have been found to contain large-sized pollen grains, cells, leaves, stomata, xylem, etc. The polyploid plants are more vigorous than diploids.
2. **Physiological effect of polyploidy.** The ascorbic acid content has been reported to be higher in tetraploid cabbages and tomatoes than in corresponding diploids. Likewise corn meal of a tetraploid maize seed contain 40 per cent more vitamin A than cornmeal from a diploid plant.
3. **Effect on fertility of polyploidy.** The most important effect of polyploidy is that it reduces the fertility of polyploid plants in variable degrees.
4. **Evolution through polyploidy.** Interspecific hybridization combined with polyploidy offers a mechanism whereby new species may arise suddenly in natural populations.

**B. ANEUPLOIDY**

Changes that involve parts of a chromosome set results in individuals, called **aneuploids** (Gr. *aneu* = uneven ; *ploid* = unit). Aneuploidy can be either due to the loss of one or more chromosomes (**hypoploidy**) or due to addition of one or more chromosomes to the complete chromosome set (**hyperploidy**). Hypoploidy is mainly due to the substraction (or loss) of a single chromosome, called **monosomy** (2n–1) or due to the

**Non-disjunction at first division**

second division

*n* + 1

first division

*n* + 1

*n* – 1

*n* – 1

**Non-disjunction at second division**

*n* + 1

*n* – 1

*n*

*n*

**Fig. 15.10.** The mode of origin of aneuploid gametes by non- dis- junction at either the first or second meiotic division (after Suzuki *et al.* 1986).

loss of one pair of chromosome called **nullisomy** (2n–2 ; two lost chromosomes are homologs). Likewise, hyperploidy may in- volve addition of either a single chromosome, called **trisomy** (2n

+ 1) or a pair of chromosomes,

called **tetrasomy** (2n + 2). In the monoploid organisms, addition of single chromosome produces **disomy** (n + 1). All of these aneu- ploids are probably produced by nondisjunction during mitosis or meiosis (Fig. 15.10).

1. **Monosomy**

Diploid organisms which are missing one chromosome of a single pair are monosomic with the genomic formula 2n –1. A monosomic individual forms ga- metes of two types, (n) and (n – 1). The n –1 gametes do not survive in plants, but, in animals that may cause genetic imbal- ance, which is manifested by high mortality or reduced fertility of resulted organism (Fig. 15.11).

**CHROMOSOMAL MUTATION II**

n

n

n–1

n–1

**Fig. 15.11.** Behaviour of a chromosome at meiosis (after Suzuki, *et al.,* 1986).

**195**

Monosomy in diploids is not tol- erated, since it creates imbalance due to loss of one complete chromosome. How- ever, in polyploids, monosomy has no apparent effect, since they have several chromosomes of same type and loss of one chromosome can be easily tolerated. The number of possible monosomics in an organism will be equal to the haploid chromosome number. For example, in common wheat, since 21 pairs of chro- mosomes are present, 21 possible monosomics are known. These 21 monosomics in wheat were produced by

**E.R. Sears** in variety called “**chinese spring**” and being used for genetic studies all over the world. Monosomics were also reported in cotton (2n= 52) by **J. Endrizzi** and his coworkers and in tobacco (2n = 48) by **E.R. Clausen** and **D.R. Cameron**. Monsomics have also been produced in maize and tomato (2n = 24) despite their being diploids. **Double monosomics** (2n–1–1) or **triple monosomics** (2n–1–1–1) could also be produced in polyploids such as wheat. Double monosomic means that the chromosome number is 2n–2 like that in a nullisomic, but the missing chromosomes are non- homologous. The same explanation is applied to the triple monosomics.

1. **Nullisomy**

An organism which has lost a chromosome pair is a nullosomic. The nullosomic organism has the genomic formula (2n–2). A nullosomic diploid often does not survive, however, a nullosomic polyploid (*e.g.,* hexaploid wheat, 6*x*–2) may survive but exhibit reduced vigour and fertility.

1. **Trisomy**

Trisomics are those diploid organisms which have an extra chromosome (2n + 1). Since the extra chromosome may belong to any one of different chromosomes of a haploid complement, the number of possible trisomics will be equal to the haploid chromosome number. For example, haploid chromosome number of barley is 7, con- sequently in it seven trisomics are possible. Fur- ther, when the extra chromosome is identical to its homologs, such a trisomic is called **primary tri- somic**. There are also secondary and tertiary trisomics. While the **secondary trisomic** means that the extra chromosome should be an isochro- mosome (*i.e.,* both chromosome arms genetically similar), a **tertiary trisomic** would mean that the extra chromosome should be the product of trans-

Primary trisomic

Secondary trisomic

Tertiary trisomic

**Fig. 15.12.** Three kinds of trisomics.

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| 1 | 1 | 1 | 3 | 3 |
| 2 | 2 | 2 | 4 | 4 |
| 1 | 1 | 1 | 3 | 3 |
| 2 | 2 | 2 | 4 | 4 |
| 1 | 1 | 1 | 3 | 3 |
| 2 | 2 | 4 | 4 | 4 |

location (Fig. 15.12). Trisomics were obtained for the first time in jimson weed (*Datura stramonium*)

by **A.F. Blakeslee** and **J. Belling** (1924). Since the haploid chromosome number of this species is n=12, so here, 12 primary trisomics, 24 secondary trisomics and a large number of tertiary trisomics are possible. Most of the trisomics were identified by the size, shape and other morphological features of the fruit of jimson weed (Fig. 15.13). In barley, such a trisomic series is produced and extensively studied by **T. Tsuchiya**.

**Trisomy in humans.** In human beings, the following three syndromes have been studied :

**GENETICS, HUMAN GENETICS AND EUGENICS**

**196**

* 1. **Down’s syndrome (DS) or Trisomy-21.**

Down’s syndrome is named after the physician **J.Langdon Down** who first described this genetic defect in 1866 and it was formally called **mongolism** or **mongolian idiocy**. It is usually associated with a trisomic condition for one of the smallest human autosomes (*i.e.,* chromosome 21). It is the most common chromosomal abnormality in live births (1/650 births). There are about 50 physical characteristics shown by DS infants soon after birth. These include mild or moderate mental



|  |
| --- |
| **(a) (b)**(a) Karyotype of a Down's syndrome child. (b) Girls suffering from Down's syndrome. |

retardation; eyes that slant up and out with internal epicanthal folds; a tongue that is large, swollen and protruding ; small and under developed ears; a single palmar crease; short stature; stubby fingers; an enlarged liver and spleen. Women over 45 years of age are about twenty times more likely to give birth to a child with DS than women aged 20. Nondisjunction of chromosome pair 21 during oogenesis is the main cause of occurrence of trisomy-21. This event is found to be affected either by senescence of oocytes, virus infection, radiation damage, etc. (*e.g.,* mothers who have had infectious hepatitis prior to pregnancy may have three times more chances to give birth to DS infants). Nondisjunction of



|  |
| --- |
| NormalRolled Glossy Buckling Microcarpic Reduced PoinsettiaElongate Echinus Cocklebur Spinach Globe IIex**Fig. 15.13.** Fruit capsules of the 12 primary trisomics of *Datura stramonium,* each with its particular phenotype (after Sybenga, 1972). |

**CHROMOSOMAL MUTATION II**

Plant of *Datura stramonium.*

**197**

chromosome pair 21 during spermato- genesis can also produce child with DS, but paternal age does not seem to be associated with its incidence.

Lastly, in about 2 to 5 per cent cases, the normal chromosome number is present (2n = 46), but the extra chromo- some 21 is attached (translocated) to one of the larger autosomes (usually chromo- some 14).

* 1. **Edward’s syndrome or Tri- somy-18.** First described in 1960 by **John**

**H. Edwards** and his colleagues, **trisomy- 18** is found to contain an incidence of about 0.3 per 1000 births. It is character- ized by multiple malformations, prima- rily low-set ears; small receding lower jaw; flexed and clenched fingers; cardiac malformations; and various deformaties of skull, face and feet. Harelip and cleft

palate often occurs. Death takes place around 3 to 4 months of age. Trisomy-18 children show evidence of severe mental retardation, which is more pronounced in females (the reason is still not clear). Like the Down’s syndrome, occurrence of Edward’s syndrome is too related with maternal age (*i.e.,* 35 to 45 year old mothers have more chance of giving birth to trisomy-18 infant).

* 1. **Patau syndrome or Trisomy-13.** This syndrome was described in 1960 by **Klaus Patau** and coworkers. Its incidence is about 0.2 per 1000 births. Individuals with Patau syndrome appear to be markedly mentally retarded; have sloping forehead, harelip and cleft palate. Polydactyly (both hands and feet) is almost always present; the hands and feet are deformed. Cardiac and various internal defects (of kidney, colon, small intestine) are common. Death usually occurs within hours or days, but the foetus may abort spontaneously. (**Note** : Sex chromosomal variations will be described in Chapter 18 of Human Genetics).



|  |
| --- |
| (a) The karyotype of Edwards syndrome. (b) The karyotype of an infant with Patau syndrome. |

**Trisomy in non-humans. Trisomy-22** has been reported in chimpanzees (**McClure** *et al.,* 1969); this shows Down syndrome-like phenotypic features. **Trisomy-21** has been reported in the gorilla.

**GENETICS, HUMAN GENETICS AND EUGENICS**

**198**

**Cytology of trisomics.** The trisomics have an extra chromosome which is homologous to one chromosome of the diploid complement. Therefore, it forms **trivalent** which may take a variety of shapes in primary and secondary trisomics (Fig 15.14). In a tertiary trisomic a characteristic pentavalent is observed.

|  |
| --- |
| Type of Somatic Metaphase I configurations trisomic chromosomes1 1 1 1 1 1 1 1 1 11. Primary 1 1 1 2 2 2

trisomics 2 2 2 2 2 22 2 2 2 2 21a b c d1 1 1 1 1 1 1 1 1 1 12 2 22 2 11. Secondary or 2 2 2

trisomics 1 1 2 2 2 2 2 2 2 2 22 22 2 2 a b c d1 1 1 3 3 2 1 4 33. Tertiary 2 2 4 4 4 2 1 1 4 4 3trisomics (a pentavalent)**Fig. 15.14.** Meiotic configurations formed at metaphase I in different types of trisomics. |

1. **Double Trisomy**

In a diploid organism when two different chromosomes are represented in triplicate, the double trisomic is resulted. The double trisomic causes great genetic imbalance and has the genomic formula 2n+1+1.

1. **Tetrasomy**

The diploid organisms having two extra chromosomes are known as **tetrasomic**. They have the genomic formula 2n+2. All the 21 possible tetrasomics are available in wheat.

Different types of ploidies can be summarized in the following chart :

Variation in chromosome number (ploidy)

Euploidy

Aneuploidy

Monoploidy Diploidy Polyploidy (n) (2n) (3n, 4n, 5n,

6n, etc.)

Hypoploidy

Hyperploidy

Monosomy

(2n–1)

Nullisomy

(2n–2)

Trisomy

(2n+1)

Double trisomy Tetrasomy

(2n + 1 +1) (2n+2)