**CHAPTER-13**

**Numerical Changes in Chromosomes**

**Abdul Rauf Shakoori1, Saira Aftab1 and Farah Rauf Shakoori2**

1School of Biological Sciences, University of the Punjab, Quaid-i-Azam, Campus, Lahore 54590, Pakistan

Email: arshakoori.sbs@pu.edu.pk, saira3aftab@gmail.com

2Department of Zoology, University of the Punjab Quaid-i-Azam, Campus, Lahore 54590, Pakistan

Email: farah.shakoori.zool@pu.edu.pk

**ABSTRACT:** Every organism has basic specific number of chromosomes, which are constant for a species. Changes in the chromosomal number, however, do occur which reflect high inviability and phenotypic anomalies. Abnormal euploidy will result if whole set of chromosome is involved and aneuploidy will result if parts of chromosomal set are involved. The most common abnormal euploids are polyploids such a triploids, tetraploids etc. Allopolyploids can be made by crossing and doubling progeny chromosomes with colchicines. This technique has important applications in crop breeding. Aneuploids result in an unbalanced genotype with an abnormal phenotype . Monosomy (2n-1) and trisomy (2n+1) are examples of aneuploid. Aneuploid conditions such as Down’s syndrome, Klinefelter’s syndrome, Turner’s syndrome are well studied in human. It is believed to be due to chromosomal nondisjunction, and constitutes major portion of genetically based ill health in human population.

**KEY WORDS:** Euploid, aneuploid, polyploid, allopolyploidy, monoploid, chromosomal nondisjunction, Down Syndrome, Klinefelter Syndrome, Turner Syndrome, Trisomy 8, Trisomy 13, Trisomy 18.

**13.1. CHROMOSOME NUMBER**

All organisms contain specific number of chromosomes, which is constant for a particular species. The number varies from two (one pair) in Jack jumper ant (*Myrmecia pilosula*) and round worm (*Ascaris lumbricoides*) to as many as 308 (154 pairs) in Black Mulberry (*Morus nigra*) and 268 (134 pairs) in butterfly (*Agrodiaetus shahrami*). Mosquito (*Aedes aegypti*) has 6 (3 pairs), chimpanzee 48 (24 pairs) and man (*Homo sapiens*) 46 (23 pairs) of chromosomes (Table 13-1).

The most commonly present chromosomal number or set of chromosomes is called euploidy number. Human has 46 chromosomes as diploid (2n) number in somatic cells and its gametes (sperms and eggs) have 23 chromosomes which is haploid (n) number. The original set of chromosomes is called monoploid and its presence in nature as duplicated form is diploidy. Aneuploidy is the condition in which animal has extra number of chromosome other than normal euploid chromosomes e.g. trisomy in humans (46+1=47) or absence of one (46-1=45) or more chromosome from the euploid set of chromosomes are examples of aneuploidy. Aneuploidy generally results from non-disjunction of chromosome and chromosomal loss during the cell division. Non-disjunction occurs when one out of two daughter cells receives an extra chromosome in mitosis (2n+1) or in meiosis (n+1) as a result of non-disjunction of homologous chromosome. The other daughter cell or gamete undergoes loss of chromosome or nullisomy. In some

**Table 13-I. Organisms (Plants and animals) listed according to the diploid number of chromosomes (2n).**

|  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- |
| No | **Name of the organisms** | **2n**  | **No**  | **Name of the organisms** | **2n** |
| 1 | Ciliate:*(Tetrahymena thermophila)*in micronucleus | 10 | 12 | Asiatic black bear*(Ursus arctos)* | 74 |
| 2 | Black mulberry*(Morus nigra)* | 308 | 13 | Black nightshade*(Solanum nigrum)* | 72 |
| 3 | Butterfly*(Agrodiaetus shahrami)* | 268 | 14 | Elk*(Cervus canadensis)* | 68 |
| 4 | Rattlesnake fern*(Botrypus virginianus)* | 184 | 15 | Chinchilla*(Chinchilla lanigera)*Horse*(Equus ferus cabalius)* | 64 |
| 5 | Northern Lamprey | 174 | 16 | Donkey*(Equus africanus asinus)* | 62 |
| 6 | Carp | 104 | 17 | American bison*(Bison bison)* | 60 |
| 7 | Red Viscacha rat*(Tympanoctomys barrerae)* | 102 | 18 | Elephant | 56 |
| 8  | Shrimp*(Penaeus semisulcatus)* | 86-92 | 19 | Sheep*(Ovis orientalis aries)* | 54 |
| 9 | African Hedgehog | 90 | 20 | Cotton*(Gossypium anatinus)* | 52 |
| 10 | PigeonTurkey | 80 | 21 | Zebra fish*(Danio rerio)* | 50 |
| 11 | Chicken*(Gallus gallus domesticus)* | 78 | 22 | Chimpanzee*(Pan trtoglodytes)*GorillaPotato*(Solanum tuberosum)*Water Buffalo*(Bubalus bubalis)* | 48 |
| 23 | Human*(Homo sapiens)* | 46 | 32 | Cannabis*(Cannabis sativa)*Maize*(Zea mays)* | 20 |
| 24 | DolphinRabbit*(Oryctolagus cuniculus)* | 44 | 33 | Cabbage*(Brassica oleracea)* | 18 |
| 25 | Rat*(Rattus norvegicus)*Rhesus monkey*(Macaca mulatta)*Wheat*(Triticum aestivum)* | 42 | 34 | Kangaroo | 16 |
| 26 | Mango*(Mangifera indica)*Mouse*(Mus musculus)* | 40 | 35 | Pea*(Pisum sativum)* | 14 |
| 27 | Cat*(Felis sivestris catus)*Lion*(Panthera keo)* | 38 | 36 | Nematode*(Caenorhabditis elegans)* | 12/11 |
| 28 | Earthworm*(Lumbricus terrestris)* | 36 | 37 | Fruit fly*(Drosophila melanogaster)* | 8 |
| 29 | Porcupine | 34 | 38 | Mosquito*(Aedes aegypti)* | 6 |
| 30  | Honeybee*(Apis mellifera)* | 32 | 39 | Australian Daisy*(Brachyscome dichromosomatica)*Jackjumper ant*(Myrmecia pilosula)*Round Worm(*Ascaris lulmbricoides*) | 2 |
| 31 | Rice*(Oryza sativa)* | 24 |

https://en.wikipedia.org/wiki/List\_of\_organisms\_by\_chromosome\_count

organisms the whole euploid set of chromosomes is repeated several times (polyploidy) making new organism triploid (3n) or tetraploid (4n) (Fig. 13-1).

**13.2. ABNORMAL CHROMOSOMAL NUMBER / ANEUPLOIDY**

Changing in chromosome number are usually classified into those changes involving whole chromosome sets and those involving parts of chromosome sets. Any change in chromosomal number leading to disturbance of naturally occurring balance of chromosome is called aneuploidy. In this chapter we will discuss different forms of aneuploidy in plants and animals and these different forms could be monoploidy, polyploidy, hyperploidy or hypoploidy.

**13.2.1. MONOPLOIDY**

Monoploidy or haploidy is naturally occurring phenomenon in insects like bees, wasps and aunts which give rise to male population from unfertilized eggs (Fig. 13.2). This process in which the development of an organism occur from an egg without fertilization is known as parthenogenesis. Monoploid cells do not undergo meiosis hence the gamete production in honey bee and wasp males are produced by mitosis.

Plant breeder use monopoidy as a selective method to easily grow a plant variety containing desired features by a technique called anther culture (Fig. 13.3). In anthers culture a pollen grain destined cell may be treated with an alkaloid colchicine which inhibits the spindle fiber development and hence blocks the cell division. These individual cells which have now two sets of chromosomes can be allowed to grow into a mass known as callus in the presence of specific culture media and then this mass may be seeded to grow into mature plant (Fig. 13-4). This technique has helped to selectively raise insects and pest resistant population of plants like soybean and tobacco (Fig.13-5).

**13.2.2. POLYPLOIDS**

Polyploidy is a condition in which multiple number of natural set of chromosomes is present in the organism. Depending upon the number of chromosomal sets , the organisms are known as triploid (with 3 chromosomes), tetraploid (with 4n chromosomes), pentaploid (with 5n chromosomes) and so on (Fig. 13-1).

**13.2.2.1. POLYPLOIDY IN PLANTS**

The polyploids evolved from diploids and became established in nature by accidental somatic doubling during mitosis or irregular reductional division in which sets of chromosomes fail to separate completely to the poles in the anaphase of reproductive cell. Once polyploidy is established intercrossing among plants with different chromosome numbers may give rise to numerous chromosome combinations. Most of these are sterile, but some may be fertile.

Some plants groups have a series of chromosome numbers based on a multiple of a basic number. In the genus *Chrysanthemum* for example the basic number is 9 and species are known that have 18, 36, 54, 72 and 90 chromosomes.

 Triploids are obtained when a tetraploid (4n) and diploid (2n) are crossed resulting in population of plants contains triploid gametes (3n) which could be further raised as triploid. It could be autopolyploid (n1+n1), if the multiple sets of chromosomes are from the same species or allopolyploid (n1+n2), if the sets of chromosomes are from different though closely related species. Different varieties of plants present day have been developed by this method to accumulate favorite traits of closely related plant varieties. An autotetraploid can be developed by non-segregation of chromosomes, accidently or by treatment with colchicines during cell division.

The first classical allopolyploid was first developed in 1928 by G. Karpechenko in an attempt to make a fertile offspring in a cross between cabbage (*Brassica*) and radish (*Raphanus*). Although these two species are different but they are closely related and both have 18 chromosomes naturally which allows an efficient intercross. He was successful in raising a hybrid but the resultant plant variety produced was sterile and was unable to cross to give rise to new progeny as the 9 out of 18 chromosomes of both were too different to pair in normal cell division (Fig. 13-6). Some seeds produced from these plants later on were successful to give rise to new plant species and their chromosomal number was 36. These new variety of plants were fertile and had equal proportion of chromosomes from both species of parent plants. This type of individual is called amphidiploids.Amphidiploids of Karpechenko had roots of cabbage and radish leaves. He called his new plant variety as *Raphanobrassica.*

Using this technique new variety of wheat (*Triticale*) has been produced by intercrossing wild type wheat (*Triticum*) and rye (*Secale*). *Triticale* combines the high yield of wheat and ruggedness of rye (Figure 13-7). Bread wheat (*Triticum* *aestivum*) with 42 chromosomes was produced from a cross between *T. monococcum* (14 chromosomes) with 00goat grass (*Aegilops*) with 28 chromosomes. Likewise the hybrids of *Gossypium* show a wide range of vigorous fertility. Old world cotton has 13 pairs of large chromosomes. American cotton has 13 pairs of small chromosomes. New world cotton has 26 pairs, 13 large and 13 small. Evidently hybridization and chromosome duplication occurred somewhere in the ancestry of the New World cotton.

**13.2.2.2. POLYPLOIDY IN ANIMALS**

Polyploidy in animal kingdom is not very common and is found in lower animals like flat worms, leeches, shrimps and insects. Among insects it is common in ants, wasps, bees, beetles, flies and butterflies. It is also common in some species of fish and lizards. These animals reproduce by a method called parthenogenesis which lacks normal reduction division of meiosis and cells are divided by mitosis. The offsprings are produced by an unfertilized female. A vast majority of insects have polyploidy as a mechanism of sex differentiation. Insects have an advanced type of parthenogenesis in which meiotic division is suppressed and eggs undergo a single maturation division called apomixis. Among insects *Saga pedo* (Pallas) (Fig. 13-8A) which is a grasshopper commonly found in countries in the north of Mediterranean is a tetraploid and *Physokermes hemicryphus* (Dalman), an hemipteran insect is triploid. Many members of Lepidoptera, Diptera and Coleoptera are also polyploid.

Polyploidy is also found in some species of fish. Initially it was thought that self-fertilization is a basic requirement for polyploidy which is a common phenomenon in plants and is also found in species of fish but as a matter of fact out of many polyploidy species of fish only one has self-fertilizing ability *Kryptolebias* (=*Rivulus) marmoratus* (Fig. 13-8B). *Catostomids* family (fresh water suckers) is tetraploid. Some very common verities of fish like gold fish, minnow and carp are also tetraploid. Figure 13-8 C shows a triploid female of sunshine bass, which is bigger in size than the normal diploid female fish.

It was thought that since animals need to have their sex determination system based on chromosome type instead of number so there are less polyploids in animal kingdom as compared to plants but now new data on chromosomal studies suggests that polyploidy has played important role in species selection and speciation in animal kingdom as well.

Among reptiles only some varieties of lizards are known to be naturally polyploids and they reproduce by parthenogenetically (Fig. 13-9). Compared to reptile, amphibians have variety of polyploids. Urodeles have triploid females which are supposed to reproduce gynogenetically, a process in which a sperm is donated by male of related bisexual and diploid species and this sperm activates the egg (Fig. 13-9). Members of *Xenopus* may be tetraploid (4x) like *X. vestitus* with 72 chromosomes, and hexaploid (6x) e.g. *X. ruwenzoriensis* with 108 chromosomes. Members of anuran family Leptodactylidae like *Ceratophrys dorsata* and *C. ornata* are octaploids (8n=104, ancestral number is 13).

Meiosis was observed in females of triploid unisexual salamander *Ambystoma tremblayi* and in triploid lizard *Cnemidophorus (=Aspidoscelis) uniparens*. They have 3 sets of chromosomes instead of normal 2. The male’s sperm only stimulates egg development and it does not contribute its genetic material. It was found that in initial prophase hexaploid oogonial cells are produced and these oogonial cells turn into triploid eggs after reduction division of meisosis (Lewis, 2012).

The fertile hybrids of European water frogs reproduce by hybridogenesis. This is a form of [reproduction](https://en.wiktionary.org/wiki/reproduction) resembling [parthenogenesis](https://en.wiktionary.org/wiki/parthenogenesis), but [hemiclonal](https://en.wiktionary.org/wiki/hemiclonal) rather than completely [asexual](https://en.wiktionary.org/wiki/asexual). Half the [genome](https://en.wiktionary.org/wiki/genome) is passed intact to the next [generation](https://en.wiktionary.org/wiki/generation), while the other half is discarded. It occurs in some animals that are [hybrids](https://en.wiktionary.org/wiki/hybrid) between species.

**13. 2.3. ANEUPLOIDY IN HUMANS**

The exact number of chromosomes in human discovered in 1956 by a group of Swedish scientists is 46 which includes 21 pairs of autosomes and two sex chromosomes (Fig. 13-10). Each human chromosome contains two arms named “p” for petite which means “small” in French and “q” for long arm and letter “q” is chosen just because it’s next to “p” alphabetically. Both long and short arms are connected to each other through centromere. Both small and long arms have different banding pattern which is important from cytogenetic point of view.

Aneuploidy occurs when spindle fibers fail to attach to any of the chromosome due to non-disjunction which in humans is mostly lethal condition. According a report by American College of Obstetricians and Gynecologists (2005) almost 1 out of 150 babies are born with chromosomal abnormalities resulting in mild to severe physical defect including mental retardation. Babies born with abnormal chromosomes have either too many or too chromosomes. Nothing can be done during pregnancies to prevent chromosomal abnormality as it is caused by the fusion of abnormal gametes either from mother or from father. According to this report almost 75% of abortions are caused by abnormal number of chromosomes in fetus. Most of the changes in chromosomal number or set of chromosomes are lethal and result in abortion or still birth. Only some changes in human chromosomes result in live births, though the newborn suffers from serious illnesses and different anomalies designated as syndrome. One of the most common aberrations in human are trisomies (2n+1) in which one chromosome is extra in genome making 47 chromosomes.

 **13.2.3.1.TRISOMY (2n+1)**

Trisomy is a condition in which an organism or a cell has one chromosome in excess of the normal somatic complement of the species. One chromosome has three homologous in place of the normal two. It is designated by 2n+1. Trisomy in humans results in mental retardation of newborn, physical abnormalities and in some carcinomas as well. Generally trisomy in humans is divided into two main types, trisomy in autosomes and trisomy of sex chromosomes. We will discuss both types one by one.

**13.2.3.1.1. TRISOMY IN AUTOSOMES**

Most commonly known autosomal trisomies of chromosome number 21 (Down syndrome), 18 (Edward syndrome), 13 (Patau syndrome) and 8 chromosome are known (Table 13-II).

**TABLE 13.II Aneuploidy Resulting from Nondisjunction in the Human Population**

|  |
| --- |
| **Chromosome Chromosome Clinical Estimated****nomenclature formula syndrome frequency** **at birth Main phenotypic characteristics** |
|   47,+21 2n+1 Down 1/700 Short broad hands with simian-type palmar Crease, short stature, hyperflexibility of  Joints, mental retardation, broad head with  Round face, open mouth with large tongue, Epicanthal fold. |
|   47,+13 2n+1 Trisomy-13 1/20,000 Mental deficiency and deafness, minor Muscle seizures, cleft lip and/or palate, Polydactyly, cardiac anomalies, posterior Heel prominence. |
|   47,+18 2n+1 Trisomy-18 1/8000 Multiple congenital malformation of many  Organs; low-set, malformed ears; receding  Mandible, small mouth and nose with  General elfin appearance; mental deficiency  Horseshoe or double kidney; short stemum. 90% die in the first 6 months. |
|  45,x 2n-1 Turner 1/2500 female Female with retarded sexual development, births usually sterile, short stature, webbing of skin In neck region, cardiovascular abnormalities, Hearing impairment. |
|   47,XXY 2n+1 Klinefelter 1/500 male Male, subfertile with small testes, developed 48,XXXY 2n+2 briths breasts, feminine pitched voice, long limbs, 48,XXYY 2n+2 knock knees, rambling talkativeness. 49,XXXXY 2n+3 50,XXXXXY 2n+4 |
|   47,XXX 2n+1 Triple X 1/700 Female with usually normal genitalia and Limited fertility. Slight mental retardation. |

**13.2.3.1.1.1. TRISOMY 21 (DOWN SYNDROME)**

Down syndrome or trisomy 21 formerly known as Mongolism (due to facial features resembling Mongolian people) is now designated as Down syndrome after Langdon Down who first described the clinical signs in 1866. Jerome Lejeune, a French pediatrician discovered the link of Down syndrome with chromosomal abnormality in 1958 (Fig. 13-11).

Since Down syndrome causes mental retardation, it was considered as the sole reason of mental retardation in humans for a long time . Other abnormalities associated to Down syndrome are dementia, hypothyroidism, congenital heart diseases and leukemia (National birth defects prevention network, 2000). Children with Down syndrome have more digestive problems compared to normal children like gastroesophageal reflux or celiac disease, swallowing problems and bowl blockage. These children also experience frequent colds, sinuses and some infections of ear and poor muscular tone leading to various physical defects shown in Table 13-III.

**Table 13-III. Anomalies in Down’s syndrome**

|  |  |
| --- | --- |
| **Anomalies in systems** | **Defects** |
| Cardiovascular system anomalies | Defects in valves |
| Hypo plastic ventricles |
| Anomalies of aortic arteries |
| Anomalies of great veins |
| Single Umbilical artery |
| Respiratory system anomalies | Anomalies of bronchus, trachea and larynx |
| Anomalies of lungs |  |
| Anomalies of digestive system | Swallowing problems  |
| Gastroesophageal reflux |
| Bowl blockage |
| Urinary system anomalies | Obstructive defects of ureter, urethra and bladder |
| Eyes | Cataracts  |
| Muscular and skeletal defects | Clubfoot  |
| Polydactyly |
| Syndactyly  |
| Nervous system anomalies | Mental retardation |
| Anomalies of genital organs | Undescended testicles |
| Hypospadias and epispadias |

Data modified from Torfs and Christianson (1998).

Presence of just one extra chromosome (Fig. 13-12) causes several abnormalities related to cardiac, respiratory and digestive systems. A list of some malformations observed in Down syndrome is given below in Table 13-III.

According to Parker et al. (2010) almost 1 out of 700 babies born has Down syndrome only in Europe. The remaining reflect fetal loss due to spontaneous abortion. This can be attributed to defect in the maternal and paternal gametes (Fig. 13-13). Recent studies have linked trisomy 21 is to mother’s age. Chances of women having a Down Syndrome child at age of 30 are 1/1000 and probability increase as the age increases e.g. at the age of 35 its 1/400 and at 40 its 1/100. A small number of people have Down Syndrome due to translocation of a piece of chromosome 21 to some other chromosome instead of complete trisomy e.g. “21q21q Robertsonian translocation”.

Human chromosome 21 is 33.5Mb long and only its small portion codes for protein. It contains 127 known genes, 98 predicted genes and 59 pseudogenes. Recent research has enabled scientist to discover Down syndrome causing loci on chromosome 21 and now different genes are known to cause different abnormalities in Down syndrome (Hattori et al., 2000). Table 13-IV below shows different genes known to cause different abnormalities in Down syndrome.

**Table 13-IV. Genes related to physical defects in Down syndrome**

|  |  |
| --- | --- |
| **Defects** | **Genes involved** |
| Defects in learning, memory and brain developmental defects | *dscam,dyrk1a, sim2, app, synj1* |
| Neurodegenerative disorders | *app, dyrk1a* |
| Motor control | *app, dyrk1a, synj1* |
| Cardiac defects | *dscam, slc19a1, col6a1* |
| Leukemia | *ets2, erg* |
| Craniofacial alterations | *ets2* |

Data modifies from Lana-Elola et al (2011).

Recent studies have shown that patients with Down syndrome have reduced chances of solid tumors which could be either due to extra dosage of tumor suppressor gene Ets2 on chromosome 21 (Sussan et al., 2008) or reduced angiogenesis due to increased dosage of *Erg,* *Jam2, Adamts1* and *Pttglip* (Reynold et al., 2010).

Prenatal diagnosis play important role in early detection of Down syndrome but nothing can be done to treat it. It is observed that society play important role to improve health and learning abilities of patient. It is observed that life expectancy of Down syndrome patient has increased from 25 to 50 years over last few decades due to improved medical facilities and public awareness. According to a survey, kids with Down syndrome are more loveable than normal kids and have same emotional feeling as normal do and little training has made parents to feel proud of their child (Fig. 13-14).

**13.2.3.1.1.2. TRISOMY 18 (47, +18; EDWARDS SYNDROME)**

### Trisomy 18 ( Fig. 13-15) is relatively rare trisomy as compared to Down syndrome but still it’s the second most common autosomal trisomy and has frequency of about 1 in 8000 live births. Trisomy 18 is also known as Edwards syndrome named after John Hilton Edwards who discovered it in 1960 (Fig. 13-16).

Clinical features of Edwards syndrome include defective cardiac system, some congenital abnormalities including kidney problems (Table 13-II). Usually new born with this conditions died within 6 months (Root and Carey, 1994) mostly in 90% of cases. Females are mostly affect by this condition, usually 80% which is three to four time more than the affected males (20%). Facial impairments include small head, prominent occiput or back part of the skull, small mouth, small jaw, short neck, low set and malformed ears, unusually shaped chest, short and prominent sternum, wide set nipples, crossed legs, flexed big toe, feet with rounded bottom, clenched hands with overlapping fingers, poorly developed finger nails, cleft or hole in iris and low birth weight (Luthardt and Keitges, 2001). Figure 13-17 shows the abnormalities of Edwards syndrome.

B

A

**13.2.3.1.1.3. TRISOMY 13 (47, +13)**

Trisomy 13 (Fig. 13-18) is the third most common trisomy and is caused by an additional chromosome number 13 (Fig. 13-19). It was first identified by Patau and colleagues in 1960 when he discovered an additional chromosome in patient (Smith et al., 1961). Abnormalities common in Patau syndrome were prescribed in 1956 by Thomas Bartholin due to which this syndrome is also called Bartholin-Patau syndrome.

Incident rate of Patau syndrome is 1 in 20,000 live births per year (Delatycki and Gardner, 1997). Seventy five to eighty percent of cases have trisomy. There are different cases of trisomy 13. In one case the whole chromosome 13 is additional and is called classic Patau syndrome or D trisomy 13. Sometimes a part of chromosome 13 is translocated additional through Robertsonian translocation producing symptoms of Patau syndrome. Mosaicism is the case in which some cells of the body have trisomy 13. Usually the life span of patient is short and new born dies in a few years but some cases have been reported where patient lives for as long as 11 years (Zoll et al., 1993).

Different anomalies include microcephaly, camptodactyly of both hands, wide gap between first and second toe, unicuspid heart valves. Several defects in internal genetalia have also been reported like in female patients didelphys uterus (with two vagina), small ovaries, multinucleated ova. Male patients had cryptorchidism, small and abnormal scrotum, hypospadias and many defects in brain development (Moerman et al., 1988).

**13.2.3.1.1.4. TRISOMY 8**

Trisomy 8 was first time reported in 1975 in a 2 month old boy with aneuploidy and mosacism for chromosome 8. Like other trisomies it also leads to some psychomotor retardation, bone and joint anomalies. Mild to moderate mental retardation is also prominent in patients. They have lower intellectual abilities and face difficulty in reasoning, concentration and have poor memory. Trisomy 8 is found to be closely related to leukemia. Almost 9% acute myeloid leukemia patients have complete chromosome 8 trisomy others have either mosaicism or robertsonian translocation (Mrozek et al., 2014). These patients also have number of mutations other than complete trisomy of chromosome 8. Almost 90% of these patients have at least one mutation commonly *RUNX1, ASXL1* and *FLT3* (round about 30% each). Patients older than 60 year more commonly have these mutations than younger patients (Becker et al., 2014).

**13.2.3.1.2. TRISOMIES IN SEX CHROMOSOMES**

Normal human male has one X and one Y chromosome while female has two X chromosomes. Any change in normal number of sex chromosomes results in various abnormalities including infertility, mental retardation, structural anomalies and development related problems. Trisomies in sex chromosomes are sometimes undetectable and patients may live normal life without being detected e.g. XYY in males. We will study the abnormal distribution of sex chromosomes in males and females individually.

**13.2.3.1.2.1. SEX CHROMOSOME TRISOMIES IN MALES**

**13.2.3.1.2.1.1. 47, XXY KLINEFELTER SYNDROME**

A condition in male with an extra x chromosome is called XXY or Klinefelter syndrome first reported by Klinefelter et al. in 1942 and characterized by hypogonadism. Figure 13-20 shows different forms of non-disjunction leaing to XXY karyotype of Klinefelter syndrome. According to a report in 2006 by National Institute of Child Health and Human Development, the incidence of this syndrome is 1 in 500-1000 new born baby boys. Major risk factor is mother’s age. Due to the presence of an extra x chromosome males tend to have female like appearance with large breast, very less facial and under arm hair growth and usually small testes. Patients with Klinefelter Syndrome usually have height greater than their parents, long legs and higher chances of gaining weight (Table 13-II).

A complete trisomy results in infertility but it is commonly observed that almost 50% of the Klinefelter Syndrome patients have sperms which is due to mosaicism of Klinefelter Syndrome instead of complete trisomy in which some cells have extra X chromosome while others do not. Patients with Klinefelter Syndrome have intellectual disabilities and are often misunderstood as lazy and shy people. Speech delay is very common in Klinefelter Syndrome there are many condition of Klinefelter Syndrome in which sex chromosomes are not in normal proportion like XXYY, XXXY, XXXXY and XXY/XY. Here it is important to know that xxyy is due to total non-disjunction in paternal chromosomes while XXXXY is due to total nondisjunction maternal chromosomes. As the number of X chromosome increases in Klinefelter Syndrome the condition becomes severe with infertility, severe to complete mental retardation and prominent physical abnormalities (Fraser et al., 1961).

Tartaglia et al. (2011) has described the medical problems in all these cases of Klinefelter Syndrome which includes asthma, food allergies, dental problems, cardiac malformation, congenital hip dysplasia, clubfoot, hypothyroidism, type II diabetes and hyper gonadotropic hypergonadism.

Patients with Klinefelter Syndrome are also observed to develop various types of cancers like testicular cancer, non-Hodgkin’s lymphoma, brain tumor and various tumors of digestive system. Breast cancer is not very common in men but 4% of all men having breast cancer are Klinefelter Syndrome patients (Hasel et al., 1995). Treatment includes hormone therapy to treat hypergonadism and speech therapy helps young Klinefelter Syndrome patients for speech delay. Psychiatric problems are also observed in Klinefelter Syndrome patients for which counseling and medication appears helpful.

**13.2.3.1.2.1.2. 47, XYY SYNDROME**

It is the most common trisomy in male population. Almost 1 out of every 1000 male is born with an extra y chromosome (Table 13-II). For long it was considered to be related to socially aggressive behavior but no solid evidence has been reported so far. Many studies done on criminals have reported higher number of XYY karyotype persons. The IQ level of XYY is usually below average due to which these patients experience problems in academic life but with extra help it can be overcome. Many of XYY have been known to secure professional degrees. Poor social life is very prominent among XYY people and due to which their marital life has been observed to suffer badly. Usually patients are fertile and most of them are not diagnosed throughout their lives. These people experience delayed motor development or defects in motor coordination in younger age. Speech delay is also observed and patients undergo speech therapy. Usually these patients are taller in height. No specific physical or facial abnormality has been observed so far (Leggett et al., 2010).

**13.2.3.1.2.2. TRISOMIES IN FEMALE (XXX, XXXX, XXXXX META FEMALE)**

Presence of an extra x chromosome in female is called x trisomy and it is very common sex chromosomal abnormality in female population. One in every 1000 female has this syndrome and risk increases as the maternal age increases. Most of the female with this condition live their life without being ever detected. Usually female with one extra x chromosome have fertility and normal gonadal activity but these females may have learning disabilities and lower IQ level also tend to have early menopause as compared to normal people. There are other cases of x chromosomal abnormality where 2 and three extra x chromosome are found but they have severe symptoms and serious mental retardation (Dewhurst, 1978). There are some cases where x chromosomal trisomy is found in mosaicism where some cells have an extra x chromosome while others do not. Females with mosaicism have lower fertility as compared to xxx females which are able to conceive and produce offspring but this offspring sometimes has some abnormalities while females with mosaicism usually face abortions, still births and if successful in delivering a child then new born has many genetic abnormalities (Luthardt and Keitges, 2001). Females with multiple x chromosomes have also been identified with some psychiatric problems like depression and having suicidal thoughts. These patients also face difficulty in accepting their gender as female and desire to be male (Turan et al., 2000). In some cases premature ovarian failure and gonadal abnormality is associated with autoimmune disorder and genitourinary problems (Michalak et al., 1983; Holland et al., 2001). Figure 13-21 shows some physical abnormalities associated with XXYY, XXXY, XXXYY, XXXXY and XXXXXY genotype.

**13.2.4. NULLISOMY (2n-2)**

Nullisomy is the condition in which one pair of chromosome is missing from normal set of chromosomes in cell. Deletion of complete pair of chromosome can occur in autosome as well as in sex chromosomes. This condition is very rare in humans and usually occurs due to defects in cell division.

**13.2.5. MONOSOMY (2n-1) (XO OR TURNER SYNDROME)**

A condition in which only one chromosome is missing out of 46 chromosomes and karyotype is 45 in human. Turner syndrome is an example. Phenotype of this syndrome was first pointed out by Turner in 1938 as infantilism, cubitus valgus, webbed neck, and short stature in females. Different scientists proposed different reasons for this phenotype until Charles Ford and his colleagues (1959) proposed XO model with 45 chromosomes and have loss of one x chromosome (Fig. 13-22) . Due to its discovery by Turner, it is also called Turner syndrome. Females with Turner syndrome are short statured, have short neck, mild neck webbing, low set ears, do not have breast and menstrual cycle like normal females and are infertile. They also tend to have higher risks of cardiac problems, diabetes and lower thyroid activity. Life expectancy of these patients is very low due to cardiac problems and incident rate is one in 25,000 live births in female population (Elsheikh et al., 2002).

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**GLOSSARY**

**A**

**Allele:** One of the possible mutational states of a gene, distinguished from other alleles by phenotypic effects.

**Allopolyploids:** Polyploidy hybrids in which the chromosome sets coming from two or more distinct, though related, species.

**Aneuploid:** Also known as heteroploid. An individual whose chromosome number is not an exact multiple of the haploid (monoploid) number for the species.

**Aneuploidy:** A condition in which the number of chromosomes is not an exact multiple of the haploid set. It is a condition of a cell or of an organism that has additions or deletions of whole chromosomes.

**Anther** **culture**: It is the process of formation of haploid plants from microspores (pollen) cultured individually.

**Apomixis:** In flowering plants apomixis is the asexual formation of a seed from the maternal tissues of the ovule, avoiding the processes of meiosis and fertilization, leading to embryo development.

**Atavism:** Reappearance of ancestral trait after several generations because of recessiveness of other masking effects.

**Autoimmune disorder:** An autoimmune disorder occurs when [the body’s immune system](https://www.nlm.nih.gov/medlineplus/ency/article/000821.htm) attacks and destroys healthy body tissue by mistake. There are more than 80 types of autoimmune disorders. An autoimmune disorder may result in the destruction of body tissue, abnormal growth of an organ, or changes in organ function

**Autopolyploidy:** Polyploid condition resulting from the replication of one diploid set of chromosomes.

**Autosomes:** Chromosomes other than the sex chromosomes. In humans, there are 22 pairs of autosomes.

**Autosomal set:** A combination of nonsex chromosomes consisting of one from each homologous pair in a diploid species.

**Autotetraploid:** An autopolyploid condition composed of four similar genomes. In this situation, genes with two alleles (A and a) can have five genotypic classes: AAAA (quadraplex), AAAa (triplex), Aaaa (duplex), Aaaa (simplex), and aaaa (nulliplex).

**B**

**Basic chromosome number (*X*):** The number of different chromosomes that make up a single complete set.

**Bartholin-Patau Syndrome:** A congenital syndrome of multiple abnormalities produced by trisomy of chromosome number 13. The trisomy is usually due to primary nondisjunction, occasionally to translocation and mosaicism.

**C**

**Camptodactyly** It is a medical condition that causes one or more fingers to be permanently bent. It involves fixed flexion deformity of the proximal interphalangeal joints. The fifth finger is always affected. Camptodactyly can be caused by a genetic disorder.

**Chromosomal aberration:** Any change resulting in the duplication, deletion, or rearrangement of chromosomal material.Abnormal structure or number of chromosomes includes deficiency, duplication, inversion, translocation, aneuploidy, polyploidy, or any other change from the normal pattern.

**Chromosomes loss:** A mechanism causing aneuploidyin which a particular chromatid or chromosome fails to become incorporated into either daughter cell during cell division.

**Crossing over:** The exchange of chromosomal material (parts of chromosomal arms) between homologous chromosomes by breakage and reunion. The exchange of material between nonsister chromatids during meiosis is the bases of genetic recombination.

**Cryptorchidism** : It is the absence of one or both testes from the scrotum. It is the most common birth defect of the male genitalia.

**D**

**Deficiency (deletion)** A chromosomal mutation involving the loss or deletion of chromosomal material.Deficiency heterozygotes are hemizygous for the genes located in the deleted segment; many deficiencies produce genetic effects similar to gene mutations. Deletion also occurs when a block of one or more nucleotide pairs is lost from a DNA molecule.

**Diploid:** A condition in which each chromosome exists in pairs; having two of each chromosome.

**Diploidy** The state of being [diploid](http://www.biology-online.org/dictionary/Diploid), that is having two sets of the [chromosomes](http://www.biology-online.org/dictionary/Chromosome) (and therefore two copies of [genes](http://www.biology-online.org/dictionary/Gene)), especially in [somatic cells](http://www.biology-online.org/dictionary/Somatic_cell).

**Disjunction:** The separation of chromosomes at the anaphase stage of cell division.

**Down syndrome:** An abnormal human phenotype including mental retardation, due to a trisomy of chromosome 21; more common in babies born to older mothers.

**E**

**Edwards syndrome: A**lso known as trisomy 18, is a chromosomal abnormality caused by the presence of all, or part of, an extra 18th chromosome. This genetic condition almost always results from nondisjunction during meiosis.

**Endpolyploidy:** The increase in chromosome sets that results from endomitotic replication within somatic nuclei.

**Euploid:** Cells containing only complete sets of chromosomes. An organism or cell having a chromosome number that is an exact multiple of the monoploid (x) or haploid number. Terms used to identify different levels in an euploid series are diploid, triploid, tetraploid, and so on.

**Euploidy:** The condition of a cell or organism that has one or more complete sets of chromosomes.

**G**

**Gynogenesis:** is the development in which the embryo contains only maternal chromosomes due to activation of an egg by a sperm that degenerates without fusing with the egg nucleus.

**H**

**Haploid:** Also known as monoploid. An organism or cell having only one complete set (n) of chromosomes or one genome. A single set of chromosomes present in the egg and sperm cells of animals and in the egg and pollen cells of plants.

**Haplotype:** Specific combination of linked alleles in a cluster of related genes. A contraction of the phrase “haploid genotype.”

**Homologous:** Chromosomes that occur in pairs and are generally similar in size and shape, one having come from the male parent and the other from the female parent. Such chromosomes contain the same array of genes.

**Hypergonadism:** It is a condition where there is a hyperfunction of the gonads. It can manifest as precocious puberty, and is caused by abnormally high levels of testosterone or estrogen, crucial hormones for sexual development. Anabolic steroids may also be a major cause of high androgen and/or estrogen functional activity.

**Hyperploid:** Having a chromosome number greater than but not an exact multiple of the normal euploid number.

**Hypogonadism:**  Reduction or absence of hormone secretion or other physiological activity of the gonads (testes or ovaries).It describes a diminished functional activity of the gonads. [Spermatogenesis](https://en.wikipedia.org/wiki/Spermatogenesis) and [ovulation](https://en.wikipedia.org/wiki/Ovulation) in males and females, respectively, may be impaired by hypogonadism, which, depending on the degree of severity, may result in partial or complete [infertility](https://en.wikipedia.org/wiki/Infertility).

**Hypoploidy A**ny decrease in chromosome number that involves individual chromosomes rather than entire sets, so that fewer than the normal haploid number of chromosomes characteristic of the species are present, as in Turner syndrome.

**Hypospadias:** It is a condition in which the opening of the urethra is on the underside of the penis, instead of at the tip.

**I**

**Inversion:** Rotation of a segment of a chromosome by 180o so that the genes in this segment are present in the reverse order; characteristic inversion loops are produced during meiosis in the inversion heterozygotes.

**K**

**Klinefelter’s syndrome:** Sterile human males with the *XXY* chromosome constitution; other associated symptoms as well.

**L**

**Linkage groups:** Associations of loci on the same chromosome. In a species, there are as many linkage groups as there are homologous pairs of chromosomes.

**Linkage number:** The number of times one strand of a helix coils about the other.

**M**

**Menopause**: It is also known as the climacteric. It is the time in most women's lives when [menstrual periods](https://en.wikipedia.org/wiki/Menstrual_period) stop permanently, and the woman is no longer [able to have children](https://en.wikipedia.org/wiki/Fertile) Menopause typically occurs between 45 and 55 years of age. It may also be caused by a decrease in [hormone](https://en.wikipedia.org/wiki/Hormone) production by the [ovaries](https://en.wikipedia.org/wiki/Ovaries).

**Mental retardation:** It is a condition diagnosed before age 18, usually in infancy or prior to birth, that includes below-average general intellectual function, and a lack of the skills necessary for daily living.

**Microcephaly:** Abnormal smallness of the head, a congenital condition associated with incomplete brain development. It is a rare neurological condition in which an infant's head is significantly smaller than the heads of other children of the same age and sex. Sometimes detected at birth, microcephaly usually is the result of the brain developing abnormally in the womb or not growing as it should after birth. Microcephaly can be caused by a variety of genetic and environmental factors.

**Mongolism:** It isa congenital disorder caused by having an extra 21st chromosome; results in a flat face and short stature and mental retardation.

**Mosaicism:** The condition of a being a mosaic.

**Mosaics:** Individuals or tissue having cells of two or more different genotypes. In such cases the cells originate in the same zygote.

**N**

**Nondisjunction:** Failure of disjunction or separation of homologous chromosomes in mitosis or meiosis, resulting in too many chromosomes in some daughter cells and too few in other. Examples: in meiosis, both members of a pair of chromosomes go to one pole so that the other pole does not receive either of them; in mitosis, both sister chromatids go to the same pole. It is responsible for defects such as trisomy.

**Nullisomic:** An otherwise diploid cell or organism lacking both members of a chromosome pair (chromosome formula 2n-2).

**Nullisomy:** A type of genome mutation in which a pair of chromosomes that are normally present in the genome is missing. Organisms that exhibit nullisomy are called nullisomes. Nullisomy, especially in higher animals, usually results in death. Viable nullisomes can be found among polyploid plants.

**P**

**Parthenogenesis:** Reproduction in which offspring are produced by an unfertilized female. Parthenogenesis is common in ants, bees, wasps and certain species of fish and lizards.

**Patau syndrome**: It is a syndrome caused by a chromosomal abnormality, in which some or all of the cells of the body contain extra genetic material from chromosome 13.

**Polyploid:** An organism with more than two sets of chro-mosomes (*2n* diploid) or genomes (e.g., triploid (*3n*), tetra-ploid (*4n*), pentaploid (*5n*), hexaploid (*6n)*, heptaploid (*7n*), octoploid (*8n*).

**R**

**Recombination:** The process by which offspring derive a combination of genes different from that of either parent; the generation of new allelic combinations. In higher organisms, this can occur by crossing-over.

**Robertsonian translocation:** Translocation arising from breaks at or near the centromeres of two acrocentric chromosomes. The reciprocal exchange of broken parts generates one large metacentric chromosome and one very small chromosome.

**S**

**Syndrome:** A group of symptoms that occur together and represent a particular disease.

**T**

**Tetraploid:** having a chromosome number that is four times the basic or haploid number.

**Translocation:** Change in position of a segment of a chromosome to another part of the same chromosome or to a different chromosome.

**Turner syndrome:** In human beings; individuals having *XO* chromosome constitution, being phenotypically female, but having rudimentary sexual organs and mammary glands.

**X**

**X linkage:** The pattern of inheritance resulting from genes lo-cated on the X chromosome.

**Y**

**Y linkage:** Mode of inheritance shown by genes located on the y chromosome.

 **EXPLANATION OF FIGURES**

Fig. 13-1. The original set of chromosomes is called monoploid or haploid and its presence in nature in duplicated form is known as diploid. Organisms containing more than two sets of chromosomes are polyploids. Triploid (3n) and tetraploids (4n) are shown in the figure.

Fig. 13-2. Parthenogenesis in members of Hymenoptera (ants, bees, wasps and sawflies) which produce haploid male (n=16) while the queen honey bee (n=32) and workers (n=32) are diploid and born from a fertilized egg. This mechanism of sex determination is called haplodiploidy.

(<http://www.glenn-apiaries.com/genetics.html>).

Fig. 13-3. Steps involved in anther culture.

Fig. 13-4. Cells from plant anther are cultured in agar medium and then in liquid plant culture containing specific amino acids which help the development of haploid plantlet and this plantlet may be grown in to full plant with desired traits.

 (<http://agridr.in/tnauEAgri/eagri50/GBPR111/lec20.pdf>)

Fig. 13-5. Resistant diploid plant is isolated growing number of somatic cells on agar medium with inhibitors and mutagens to create mutation. Resistant plantlet is selected on basis of desired mutation in agar medium. This plantlet may be treated with Colchicine to get a diploid plant resistant plant which is also fertile.

 (<http://agridr.in/tnauEAgri/eagri50/GBPR111/lec20.pdf>)

Fig. 13-6. Development of an amphidiploid *Raphanobrassica,* which a hybrid of radish (*Raphanus*) and cabbage *(* *Brassica*) by G. Karpechenko in 1928. Both radish and cabbage have 18 chromosomes (2n), which on crossing form sterile F1 hybrid (n+n=9+9). Some seeds of this hybrid plant develop into a fertile plant containing 36 chromosomes (2n+2n=18+18; 4n=36).

 (<http://agridr.in/tnauEAgri/eagri50/GBPR111/lec20.pdf>)

Fig. 13-7. Amphidiploid *Triticale* is developed by intercross between rye pollens and wild type of wheat. The hybrid infertile first generation may sprout on sowing, while on treatment with colchicine will develop into a mature plant. The second generation seed would be fertile. Alternatively the hybrid seed’s embryo can grow on culture medium , which on treatment with colchicine will have its chromosomes doubled, and the mature plant so developed will have second generation fertile seeds.

(<http://agridr.in/tnauEAgri/eagri50/GBPR111/lec20.pdf>).

Fig.13-8. **A,**A tetraploid grasshopper *Saga pedo* Pallas; **B,** A triploid hermaphrodite mangrove fish,  *Kryptolebias* (= *Rivulus) marmoratus;* **C,** The sunshine bass on top is a diploid female which is filled with eggs (gravid). In contrast, the female on the bottom is triploid. She is bigger, even compared to a gravid fish. The gigas effect in polyploids is real, and effects sport fishing. Everyone wants to catch a bigger fish.

(<https://sites.google.com/site/orthopteraphotos/home/saginae/saga-pedo-pallas-1771>)

(<http://www.bbc.com/earth/story/20150519-the-most-extreme-fish-on-earth>)

<https://www.google.com.pk/imgres?imgurl=http://4.bp.blogspot.com/-9-nojIwFZEs/UPRex_ipvxI/AAAAAAAABVY/VtJ9gWCq7kg/s1600/2-%252Bfish.jpg&imgrefurl=http://biologicalexceptions.blogspot.com/2013/01/carp-diem-polyploid-fish-seize-day.html&h=422&w=533&tbnid=kUJbzuLWhzKuaM:&docid=hGhHxxeduAFtMM&ei=xs5_VsG2HtOSuATJ7L2IDg&tbm=isch&ved=0ahUKEwjBsZGH-_vJAhVTCY4KHUl2D-EQMwgZKAAwAA>

Fig.13-9. Different modes of unisexual reproduction in animals. Parthenogenesis is a natural form of [asexual reproduction](https://en.wikipedia.org/wiki/Asexual_reproduction) in which growth and development of [embryos](https://en.wikipedia.org/wiki/Embryos) occur without [fertilization](https://en.wikipedia.org/wiki/Fertilization). In [animals](https://en.wikipedia.org/wiki/Animal), parthenogenesis means development of an embryo from an unfertilized [egg cell](https://en.wikipedia.org/wiki/Gametophyte) and is a component process of [apomixis](https://en.wikipedia.org/wiki/Apomixis). Gynogenesis is a special form of sexual reproduction in which insemination is necessary but the head of the sperm penetrating into the ovum does not transform into male pronucleus; and the gynogenetic embryo develops at the expense of the ovum nucleus only. Hybridogenesis is a form of [reproduction](https://en.wiktionary.org/wiki/reproduction) resembling [parthenogenesis](https://en.wiktionary.org/wiki/parthenogenesis), but [hemiclonal](https://en.wiktionary.org/wiki/hemiclonal) rather than completely [asexual](https://en.wiktionary.org/wiki/asexual): half the [genome](https://en.wiktionary.org/wiki/genome) is passed intact to the next [generation](https://en.wiktionary.org/wiki/generation), while the other half is discarded. It occurs in some animals that are [hybrids](https://en.wiktionary.org/wiki/hybrid)

(<http://alfa-img.com/show/examples-of-parthenogenesis.html>).

Fig.13-10. Normal human karyotype with 23 pairs of chromosomes with 22 autosomes and one sex chromosome pair.

 (<http://ghr.nlm.nih.gov/handbook/illustrations/normalkaryotype>)

Fig.13-11. Jerome Lejeune with Down syndrome child.

Fig.13-12. Karyotype of a female patient with Down syndrome (Luthardt and Keitges, 2001).

Fig.13-13. Probable risk factors for Down syndrome.

(Modified from Gardner *et al.,* 2004. *Chromosome abnormalities and genetic counseling).*

Fig.13-14. Down syndrome patients in sports and society (Various internet sources)

Fig.13-15. Karyotype of a child with trisomy 18 syndrome, showing three No. 18 chromosomes.

Fig.13-16. **A,** John Hilton Edwards; **B**, **C**, Patient with Edwards syndrome having all characteristic features including over-riding fingers, tracheostomy and prominent back skull (Cereda and Carey, 2012).

<https://www.google.com.pk/imgres?imgurl=https://sites.google.com/site/edwardssyndromejw/_/rsrc/1324046229871/home/edwards_syndrome.jpg%253Fheight%253D320%2526width%253D267&imgrefurl=https://sites.google.com/site/edwardssyndromejw&h=317&w=267&tbnid=fl8mr2QaVlprsM:&docid=Nqru1OCQA9c6OM&ei=pc1_VpX0MIPtugTHlJb4Bg&tbm=isch&ved=0ahUKEwjV2rz9-fvJAhWDto4KHUeKBW8QMwgvKAAwAA>

Fig.13-17. Clinical features of Edwards syndrome.

Fig.13-18. Karyotype of a newborn with trisomy 13 syndrome, showing three No. 13 chromosomes.

Fig.13-19. Phynotypic variations in trisomy 13. **A,** Cyclopia; **B,** Cebocephaly; **C,** premaxillary agenesis; **D,** typical holoprosencephaly (Images taken from original study by Moerman et al., 1988).

Fig.13. 20. Different forms of non-disjunction leading to the 47,XXY karyotype of Klinefelter’s syndrome (Lanfranco et al., 2004).

Fig.13.21. Photograph of Physical features in XXYY, XXXY and XXXXY including: (A) Fifth-digit clinodactyly (and nail biting), (B) prominent elbows with hyperextensibility and (C) lower extremities with low muscle bulk in the calves, flat feet and mild pronation at the ankles (Tartaglia et al., 2011).

Fig.13.22. karyotype of Turner Syndrome with only one sex chromosome which is x chromosome. Other counterpart of x chromosome is missing making karyotype 45, XO (Luthardt and Keitges, 2001).