CHAPTER

9

Chromosomes and their Aberrations

SN-1 Gene

Introduction: It is the hereditary unit formed by segments of DNA (deoxyribonucleic acid).

1. **Function:** It synthesizes the polypeptide.
2. **Number:** There are about 80,000 genes in a human cell.
3. **Composition**
   - A. Deoxyribose sugar,
   - B. Nitrogen bases, and
   - C. Phosphates.
4. **Parts**
   - A. The *functional* part is called *exon*.
   - B. The *silent* part is called *intron*.
5. **Position**
   - A. *Locus:* It is the position of the genes on chromosome.
   - B. It is described in relation with centromere.
   - C. Alleles (*allos*—another): Any alternative form of a gene that can occupy a particular chromosomal locus.
6. **Types**
   - A. *Regulator* gene.
     - a. It represses (prevents) the activities of genes.
     - b. It inhibits protein synthesis.
   - B. *Operative* gene.
     - a. *Site:* It is present at one end of particular gene.
     - b. *Function:* A gene that serves as a starting point for reading the genetic code and controls the activity of the structured genes by interacting with repressor.
   - C. *Dominant* gene: It *expresses* its physical or biochemical trait, when allelic genes are either homozygous or heterozygous, e.g.

a. The person showing brachydactyly, the prominent genes are NB, BB.

b. The tallness is caused by dominant gene, the genotype of the tall individual is T: T, T: t.

D. *Co-dominant gene:* When both allelic genes are dominant but of two different types, both traits may have concurrent expression.

E. *Recessive gene:* It expresses its biochemical and physical traits only in homozygous state, e.g. albinism which is recessive.

F. *Sex-linked gene*

a. Abnormal gene located on X or Y chromosome.

b. 'X' linked inheritance is more common and is mostly expressed by recessive gene.

G. *Sex limited gene:* Born by autosomes but trait is expressed in one of the sex, e.g. gout, baldness.

H. *Carrier gene:* Heterozygous recessive gene acts as carrier gene. It may be expressed in subsequent generation.

SN-2 Barr body (sex chromatin)

Introduction: It is an inactivated X chromosome attached to nuclear membrane. It is found by Barr and Bertram in 1949.

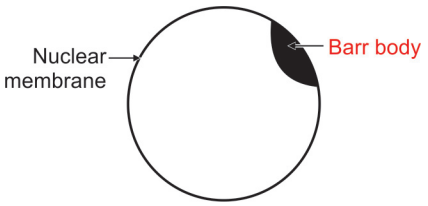


Fig. 9.1: Barr body

1. **Morphology**

A. **Site:** It is attached to nuclear membrane.

B. **Nature:** Heterochromatin (State of chromatin in which it is dark staining and tightly coiled, forming irregular clump or Barr bodies in the nuclei of cells in interphase.)

C. **Shape:** Planoconvex 

D. **Dimension:** 1 μ

E. **Staining:** Darkly stained

F. In female ♀ ⟶ XX

G. In male ♂ ⟶ (XY). There is only one X chromosome which is for cellular function. Y chromosome is for the determination of sex. Hence, there is no Barr body.

2. Person with XO will be female ♀ as the Y chromosome is absent. The only X chromosome that is present will be in an extended stage and no Barr body is seen.
   A. The inert X chromosome (Barr body) also divides during cell division. This Barr body is also known as sex chromatin.
      a. Cell + Barr body = Chromatin positive
      b. Cell – Barr body = Chromatin negative
3. **Appearance:** It appears by 2nd week of gestation.
4. **Lyon's hypothesis:** The number of Barr bodies is less than the total number of X chromosome.
   Number of X chromosome – 1 = Number of Barr bodies, e.g.
   A. In female ♀ with XX chromosome, there is a Barr body.
   B. In male ♂ with XY chromosome, there is no Barr body.
   The absence of Barr body is not enough to prove that cell is from a male ♂.

Table 9.1: Number of **B**arr bodies in different syndromes

Name of chromosomes	Number of Barr bodies	Syndrome
• XX	• One	–
• XY	• No Barr body	–
• XXX	• 2	• Triple XXX
• XXY	• 1	• Klinefelter's
• X	• 0	• Turner's
• XO	–	

5. **Applied anatomy**
   - ➢ It is helpful for the determination of sex in case of genital ambiguity.
   - ➢ It is a supplementary test in chromosomal aberrations.
   - ➢ It is helpful for diagnosis of various syndromes.

SN-3 Structure of chromosome

1. All chromosomes consist of two parallel identical filaments called chromatids.
2. They are joined together at narrowed constriction called *primary constriction*. Inside the primary constriction, there is a pale staining region called *centromere*.
3. Free ends of chromatids are called *telomeres*. Each chromatid is divided by centromere in two arms.
4. In certain chromosomes, there is another narrowing (constriction) near 1 end of each chromatid called *secondary constriction*. It is stained faintly. If secondary constriction is close to telomere, then the terminal knob of chromosome is termed as *satellite (companion)*. As per the position of centromere, chromosomes are grouped in 4 types (Table 9.2 and Fig. 9.2).

Table 9.2: Types and position of chromosome

Type	Position of centromere in chromosome
• Metacentric	• Middle
• Submetacentric	• Between midpoint and end of the chromosome
• Acrocentric	• Near one end
• Telocentric	• At one end not seen in humans

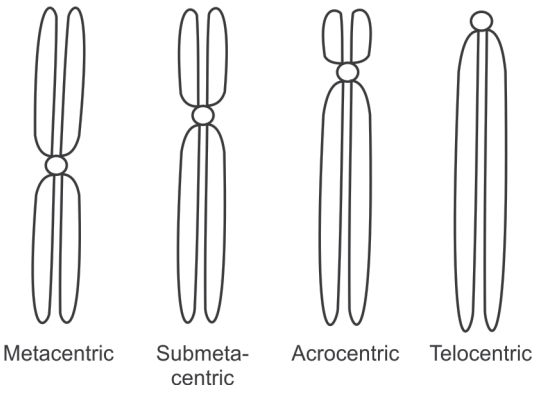


Fig. 9.2: Types of chromosomes

5. The chromosome arranged in descending order of length. The pair *No. 1* is the *longest* and the pair *No. 22* is the *shortest*. They are grouped into 7 groups. They are denoted as A to G.

Table 9.3: Classes of the chromosome

Group	Chromosome number	Features
• A	• 1 to 3	• Large, metacentric
• B	• 4 and 5	• Large, submetacentric
• C	• 6 to 12	• Medium sized, submetacentric
• D	• 13 to 15	• Medium sized acrocentric with satellite
• E	• 16 to 18	• Short submetacentric
• F	• 19 and 20	• Short metacentric
• G	• 21 and 22	• Very short acrocentric

6. Sex chromosomes are *X and Y, X belongs to C* group and *Y belongs to G* group.
7. Each cell contains fixed number of chromosomes, which is characteristic of that species or organism.
8. In somatic cell (body cell) of man, the number is 46, which is diploid number.
9. In germ cell, i.e. ova and sperms, the number is 23, called haploid number. When fertilization takes place, union of two haploid cells restores diploid number of fertilized ovum. The number of sets is termed as *ploidy*. If more than two sets are present, the cell is said to be *polyploid*. Chromosomes are in multiples of haploid number that is *tetraploid* (*tetra*—4) it has 4 times *haploid* number of chromosomes. *Triploid* (*tri*—3) number of chromosomes will be 69, i.e. 3 times the haploid number.

SN-4 Classification of chromosomes

(*Chrom*—colour, *soma*—body)

1. **Structure:** Each chromosome is made up of two identical parallel filaments called *chromatids*, which are held together at a narrow-constricted region, usually pale staining, known as *primary constriction*, or *centromere* or *kinetochore*. This structure is visible only during *metaphase* stage of cell division.
   A. Chromosome consists of
      a. **Centromere:** The constricted part in chromosome is called centromere.
      b. **Telomere:** Free ends (arms) of chromosome.
      c. **Satellite** body: Part distal to secondary construction.
2. **Function of chromosome** is for perpetuation of species.
3. **Classification**
   A. *According to functions*
      a. *Autosomes:* 22 pairs in human.
      b. Sex chromosomes decides the sex of a person.
         I. Male ♂—XY
         II. Female ♀—XX
   B. According to the positions of the centromere (*Denver's* classification).

Table 9.4: Positions of the centromere

No.	Particulars	Metacentric	Submetacentric	Acrocentric	Telocentric
1.	Centromere	Centrally	Subcentrally	Near one end	At one end so that chromatid has only one arm.
2.	Arms	Equal	p arm (short) q arm (long)	p arm shortest q arm long	One side arms
3.	Satellite body	–	–	May present	–
4.	Secondary construction	–	–	May present	–
5.	Remark	–	–	–	Not found in human being
6.	Chromatid				Only one arm

4. **Applied anatomy:** Chromosomes are mapped according to length of arm and position of centromere and it is called karyotyping.

SN-5 Chromosomal aberrations

Introduction: It is the change in the structural component of the chromosome or number of chromosome. The deletion of a segment or addition of a segment from other chromosomes results in structural aberration. The change in number leads to numerical aberration.

1. **Factors:** Following are the factors for the chromosomal variations.
   A. Late age of parents for conception.

B. Genes predisposing to non-disjunction.
C. Viral infection during pregnancy.
D. Exposure to radiation.
E. Autoimmune disease of parents.

2. **Types**

A. **Numerical** type
   a. **Aneuploidy:** 2n + 1, 2n – 1
   b. **Polyploidy:** Multiple of n except 2n, e.g. triploidy.

B. **Structural type:** It causes change in the number or sequence of genes.
   a. **Inversion:** It is the chromosomal aberration. It is caused by inverted reunion of a chromosome segment after breakage of a chromosome at 2 points. It results in a change in sequence of genes or nucleotides, e.g. the sequence *mnopq* may be inverted to *mnqop*. It may be
      I. **Paracentric:** It is on one side of centromere.
      II. **Pericentric:** It is surrounding the centromere.
   b. **Deletion** (Fig. 9.3): A portion of chromosome is lost, e.g. cri du chat syndrome or Philadelphia chromosome. It may be
      I. **Termination**–*Interstitial*

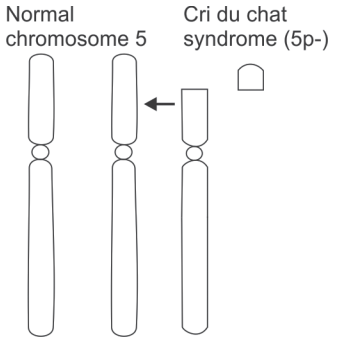


Fig. 9.3: Deletion

   c. **Translocation:** It is displacement of a portion of one chromosome to another chromosome. It is of two types
      I. Robertsonian translocation (Fig. 9.4), and
      II. Reciprocal translocation.

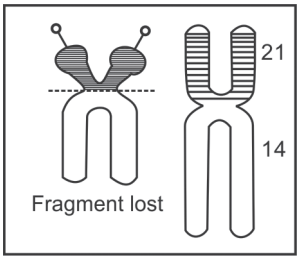


Fig. 9.4: Robertsonian translocation

d. Insertion

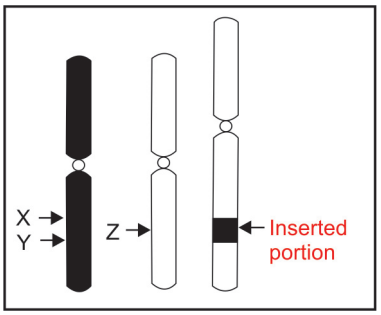


Fig. 9.5: Insertion

e. Ring chromosome

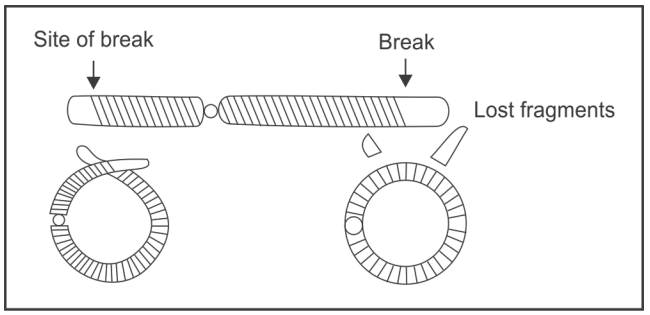


Fig. 9.6: Ring chromosome

f. Isochromosome
g. Duplication

SN-6 Chromosome banding

Introduction: The chromosomes are identified by banding technique.

1. **Procedure:** The chromosomes are treated 1st with trypsin and then stained. All the chromosomes are stained with dark and light regions. The dark regions are known as *bands*. Position of bands in normal chromosome remains fixed and is different for different chromosomes.
2. **Types of banding:** Banding can be of different types.
   A. GTG = Giemsa Trypsin Giemsa banding.
   B. ASG = Acetic saline Giemsa banding.
   C. Q = Quinacrine mustard banding.
3. **Importance of banding:** It helps in
   A. Identification of individual chromosome.
   B. Confirmation of deletion and inversion.

SN-7 Trisomy 21

Introduction: It is **most common autosomal abnormality syndrome** described by John Langdon Haydon Down 1866.

1. **Genotype:** Trisomy 21 (Down's syndrome, Mongolism) 47 XX (+ 21) or 47 XY (+21).
2. **Phenotype:** It is most common autosomal abnormality.
3. **Incidence:** 1:650 to 700 newborn.
4. **Symptoms and pathophysiology:** There are three chromosomes in chromosome no. 21.
   - A. *Forms of Down syndrome:* There are 3 forms
     - a. Trisomy 21 is most common chromosomal abnormality: Extra copy of chromosome in 21 no. of chromosome. It may be male ♂ or female ♀.
       - I. If there is XY in 23 no. of chromosome, it is male ♂.
       - II. If there is XX in 23 no. of chromosome, it is female ♀.
     - b. *Translocation Down syndrome:* It is less common. It affects 3% of people.
     - c. *Mosaic Down syndrome:* It affects 2% of people.
   - B. Cause is not known.
   - C. It is the result of
     - a. *Non-disjunction:* In 95% cases, Down syndrome is the result of non-disjunction. Here chromosomes do not split apart. This can occur in 1st or 2nd stage.
     - b. *Robertson's translocation*: In 4% of cases, Down syndrome is the result of Robertson translocation. A chromosome number 21 gets attached to chromosome number 14. An abnormal chromosome is called 14, 21. Here number of chromosomes are 46.
     - c. *Mosaic:* 1%—cells mixed.
5. **Risk factors**
   - A. **Major risk factors:** *Maternal* age
     - a. The incidence of Down syndrome is *1:1500* in females ♀ who are less than *20* years old.
     - b. The incidence of Down syndrome is *1:25* in females ♀ who are more than *45* years old.
     - c. How the maternal age affects the disjunction: In females ♀, all the X chromosomes are formed before birth. *As the maternal age advances, the X chromosome also ages. And the frequency of non-disjunction in meiosis increases with age. The number of aneuploidy cells increases with maternal age.*
   - B. **Minor risk factors** *for chromosomal variations are*
     - a. Genes predisposing to *non-disjunction.*
     - b. Viral infection during pregnancy.

c. Exposure to radiation.

d. Autoimmune disease of parents.

6. **Clinical features**

A. Effect on *brain*

B. Affected individual is mentally retarded.

C. He may suffer by Alzheimer's disease.

7. **Effect on face**

A. Flat facial profile

B. Nasal bridge is flat.

C. Palpebral fissure is slanting upwards at lateral end.

D. Epicanthic fold of eyes.

E. Maxilla is small.

F. Palate is narrow, so the oral cavity cannot accommodate tongue.

G. The tongue protrudes out of mouth.

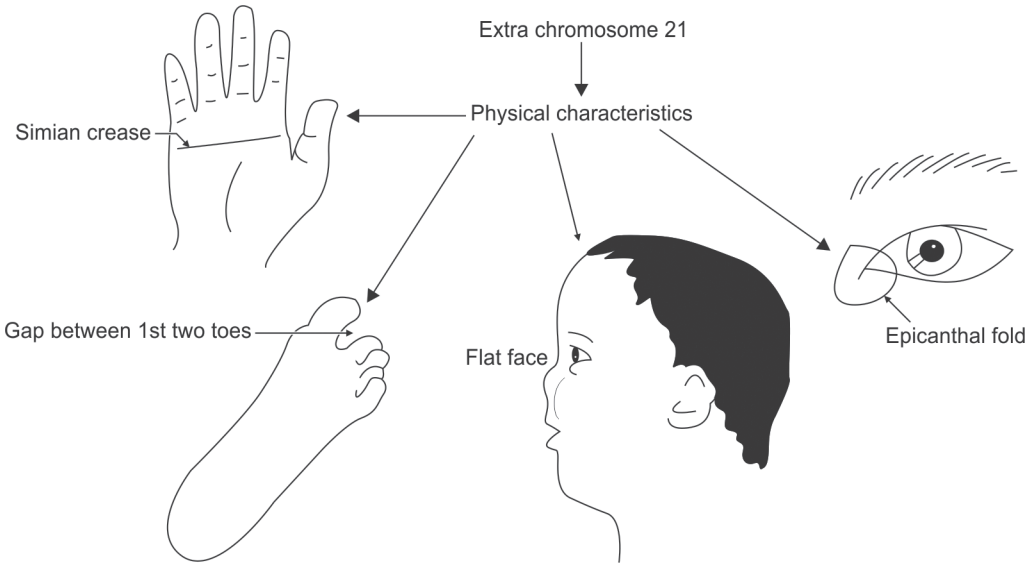


Fig. 9.7: Down syndrome (trisomy 21)

8. **Effect on heart:** In about 50% of the cases, there is congenital heart disease. There is atrioseptal defect.

9. Effect on **gastrointestinal** tract**:** Duodenal atresia

10. Effect on **blood:** Acute lymphatic leukaemia

11. **Reproductive**: Sterility in males ♂

12. **Physical characteristics**

A. Simian crease

B. Gap between two toes

13. **Investigations**
    A. Blood
        a. There is decrease of
            I. Alpha-foetoprotein
            II. Unconjugated estriol (uE3)
        b. There is increase of
            I. Human chorionic gonadotrophin (hCG)
            II. Inhibin A
    B. Ultrasound: Nuchal transparency—how much light passes through neck.

SN-8 Turner syndrome (45 X)

Introduction: Described by Turner in 1938.

1. **Monosomy** 23
    A. It is not a recessive nor dominant sex-linked disorder.
    B. It is loss of one X chromosome
    C. Incidence: 1:2000 girls
    D. **Aetiology**
        a. *Major risk factors: Maternal* age
            I. How the maternal age affects the disjunction: In females ♀, all the X chromosomes are formed before birth. *As the maternal age advances, the X chromosome also ages. And the frequency of non-disjunction in meiosis increases with age. The number of aneuploidy cells increases with maternal age.*
        b. *Minor risk factors for chromosomal variations are*
            I. Genes predisposing to *non-disjunction*.
            II. Viral infection during pregnancy.
            III. Exposure to radiation.
            IV. Autoimmune disease of parents
2. **Incidence:** 1:5000 newborns.
3. **Genotype:** 45 X
4. **Phenotype:** In female ♀, Barr body is absent.
5. **Clinical manifestations**
    A. Stature—*short female* ♀
    B. Mouth—shark-like.
    C. Lip
        a. Upper—curved.
        b. Lower—straight.

D. Chest: Shield-like—breast under developed, widely placed rudimentary nipples.
E. Genitalia: Small—ovarian dysgenesis infertility
F. *Most often sterile (infertile)*
G. *In females ♀, gonads are not developed at puberty.*
H. *Mental retardation*
I. It is a genetic disorder that only affects females ♀.
J. But girl with a Turner's syndrome has only 45 chromosomes.
K. Face more health problems than average female ♀.
L. Low intelligence.
M. Webbing of neck.

SN-9 Klinefelter syndrome (47 XXY)

Introduction: Described by Klinefelter in 1942.

1. **Interesting facts**
   A. George Washington has this syndrome.
   B. One Barr body is present.
   C. Commonest chromosomal disorders in male ♂.
   D. An extra X chromosome in male ♂.
   E. Woman who get pregnant after age of 35 are slightly more likely to have a boy with the syndrome than younger women.
   F. It is not inherited. These chromosomal changes occur as random events.
   G. Nondisjunction results in reproductive cell with an abnormal number of chromosomes.
2. **How does Klinefelter come about:** An egg or sperm cell may gain one or more extra copies of the X chromosome as a result of nondisjunction. If one of the atypical reproductive cells contributes to the genotype of a child, the child will receive an extra X.
   It is because of nondisjunction. It is failure of paired chromosome to disjoin (separate) during cell division so that both chromosomes go to one daughter cell and none to other.
3. **Incidence:** It occurs in about 1 in 500 to 1000 baby boys.1:1000 live male ♂ birth.
4. **Phenotype:** Male ♂.
5. **Genotype:** 47 XXY
6. **Signs and tests**
   A. **Karyotyping:** A test to examine chromosomes in a sample of cells, which can help identify genetic problems. This test can count the number of chromosomes, and different structures in chromosomes.
   B. **Semen** test: It is a test to measure the amount and quality of a man's semen. This test would be completed because the most common symptom is infertility.

7. **Clinical manifestations**
   A. **Abnormal body proportions:** Long legs, short trunk. Shoulder equal to the hip size.
   B. **Gynecomastia:** Abnormally large breast.
   C. **Infertility**
   D. **Sexual** problems
   E. Scanty pubic hair.
   F. Small, firm testicles.
   G. **Azoospermia**.
8. **Diagnosis**
   A. Prenatal diagnosis by chorionic sampling or amniocentesis in which fetal tissue is extracted and DNA is examined.
   B. There is no way to detect carrier of this disorder.
   C. Karyotyping
   D. Semen test
   E. Amount and quality of semen
9. **Complications**
   A. Enlarged teeth with a thinning surface (*taurodontism*)
   B. *ADHD* (attention deficit hyperactivity disorder)
   C. Breast cancer in men
   D. Depression
   E. Learning disabilities including dyslexia (disorder involving difficulty in learning to read or interpret words, letters and other symbols).
   F. **Osteoporosis:** Bones become fragile and more likely to fracture
   G. **Varicose** veins
   H. **Autoimmune** disorder
      a. Lupus
      b. Rheumatoid arthritis
      c. Sjogren syndrome
10. The extra X chromosome has many effects on the body particularly on the testis.
   A. No puberty
   B. No testosterone
   C. Size of testis is 1/8th the size.
11. **Treatment**
   A. No treatment available to change a person's chromosomal makeup.
   B. Testosterone replacement for
      a. Muscularization
      b. Positive mental attitude
      c. Less fat on abdomen
      d. Body hair
      e. Infertility

SN-10 Cri du chat syndrome (5p-)

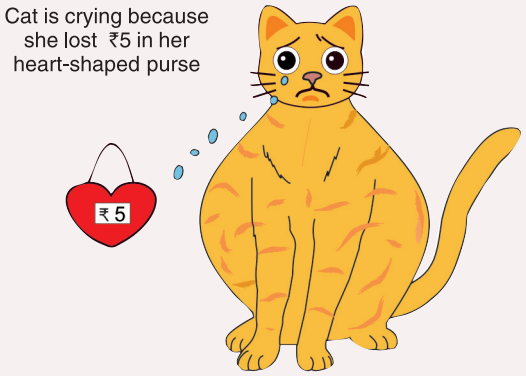
Pic Mnemonic—Tip

Box 9.1: Cri du chat syndrome

Cat is crying for losing heart shaped purse having ₹5 in it.

Illustration

1. Cat is used to denote the infant's cry which is sounding like mewing of cat.
2. Crying for ₹5 indicates low intellectual and mental retardation.
3. Heart shaped purse indicates congenital heart disease (VSD—ventral septal defect).
4. Losing ₹5 indicates deletion of small arm of chromosome no. 5
5. Picture has small head indicating microcephaly.



Introduction: *Cri du chat* (French for the cry of cat) *syndrome*. It is an example of a condition caused by structural chromosomal aberration (Fig. 9.8).

1. **Aetiology:** Genetic disorder that is caused by a missing piece of chromosome no.5
   - A. Autoimmune disease of parents.
   - B. Viral infection in pregnancy.
   - C. Conception occurred late.
   - D. Genes predisposing to nondisjunction.
   - E. Exposure of radiation.
2. **Incidence:** 1:50,000
3. **Gender variation:** Equally in male ♂ and female ♀. No specific race.
4. **Genotype:** *It is a microscopically detectable deletion of* terminal portion of short arm of p of chromosome 5 (5p-).

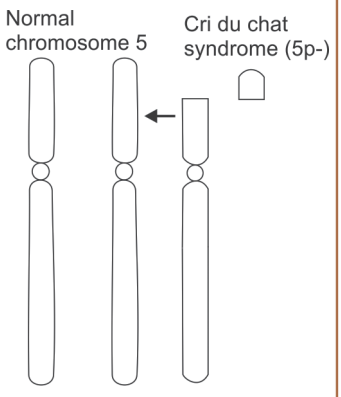


Fig. 9.8: Cri du chat syndrome

5. **Clinical manifestations**
   A. About *brain*
      a. Microcephaly
      b. Mental retardation
      c. Intellectual disability
   B. About *face*
      a. Abnormal shaped ears
      b. Premature graying of hair
      c. Skin tags just in front of the ears and eyes
      d. Wide set of eyes
      e. Oblique palpebral fissure
      f. Saddle nose
      g. Cat-like cry during infancy.
      h. Excessive drooling (dribbling of saliva)
   C. Larynx malformed, high-pitched voice
   D. *General features*
      a. Low birth weight and slow growth.
      b. Muscular hypotonia.
      c. Partial webbing or fusing of fingers or toes
      d. Single line on the palm of the hand
      e. Slow or incomplete development of motor skills
   E. *Behavioural* problem, constipation.
   F. Feeding problem
6. **Diagnosis:** Possible to detect it with amniocentesis, chorionic villus sampling or CVS
7. **Treatment:** No ways to manage the symptoms
   A. Speech therapy
   B. Physical therapy
   C. Special education
8. **Fun facts**
   A. Less noticeable as the baby gets older.
   B. Geneticist–*Jerome Lejeune identified. He also identified Down's syndrome*
   C. The main issue located in band 5P 15.2
   D. Not dominant or recessive trait
   E. 80% of the time defective chromosomes from father's side.
   F. It happens randomly.
   G. It is not hereditary.
   H. Lifespan: Normal

SN-11 Nondisjunction

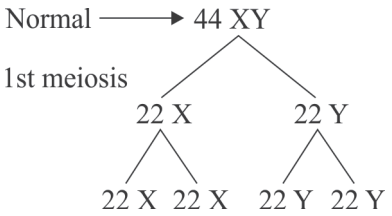
Introduction

A. Failure of normal migration of chromosome or chromosomes during *anaphase* of meiosis I.

B. Failure of migration of chromatid or chromatids during meiosis II.

1. **Reason:** Factors responsible
   A. Faulty spindle formation.
   B. Slow movement of chromatid or chromosome during anaphase.
   C. Radiation, viruses, autoimmune disease, e.g. myasthenia gravis, AIDS.
2. **It results in** formation of abnormal gametes

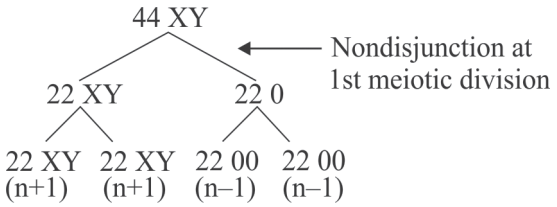
Flowchart 9.1



Normally each sperm has haploid number.

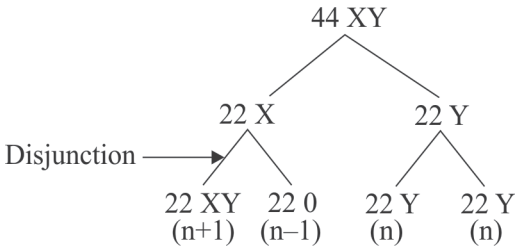
A. In nondisjunction

Flowchart 9.2



B. Nondisjunction at 2nd meiotic division

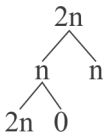
Flowchart 9.3



C. *Change in number of chromosomes:* It can occur in sex chromosomes and autosomes when (n+1) fertilized by n
→ result 2n + 1 chromosomes (trisomy)
→ (n–1) fertilized by n
(2n–1) chromosomes (monosomy)

Effect of fertilization—abnormal gamete.
In meiosis II, if all chromatids go to one side. One daughter cell with 2n and other dies

Flowchart 9.4



2n fertilizes by n ⟶ 3n

SN-12 Aneuploidy

(*An* —not, *eu*—good, *ploidy*—multiplication)
If number of chromosomes in body cell is either

1. **More than diploid number** but not multiple of haploid number, e.g. 47 chromosomes.
2. **Less than diploid number** but not haploid number, e.g.
   A. An abnormal X chromosome
   B. One more autosome goes in other cell during division
      a. Each of 46 chromosomes is a member of homologous pair. One member of each pair being received from mother, one from father. The members of pairs are called homologues. 22 pairs are similar in males ♂ and females ♀ and are called autosomes.
      b. Chromosomes in remaining pairs are sex chromosomes. In female ♀, sex chromosomes (X and X) are identical so females ♀ are *homogametes*. In males ♂, one is X and other is Y which are unequal, so males ♂ are *heterogametic*.
      c. Homologous chromosomes in each pair of autosomes are indistinguishable, i.e. two chromosomes forming pair number 5 is not identified separately as they appear same.

SN-13 Spectral karyotyping (Sky)

Karyotyping: Analysis of chromosome.

1. **Definition:** A picture of all the chromosomes in an individual cell, is arranged in homologous pairs. They are sorted according to size. Autosomal chromosomes are arranged first. Sex chromosomes are arranged last. It is also called painting of chromosomes.
2. **Spectral karyotyping:** Fluorescent tag (dye) is given. They are given by different colours.
   A. Prerequisite for spectral karyotyping
      a. Sequence of genome must be known.
      b. DNA must be in single strand.

c. Segment wise probe is made.
d. Different probes will bind different regions.
e. The designing of probe is equally important.
f. Complimentary probes are designed.
g. Depending upon various colours, we can identify the various chromosomes.
h. Hybridization of colours is possible in swapping events. It is good for
I. Translocation and
II. Substitution of chromosomes

3. **Disadvantage of spectral karyotyping:** It does not help to find other type of mutation, e.g.
A. *Inversion of chromosome:* The changes are in the same chromosome.
B. *Duplication of chromosome*

4. **Prerequisite:** The cells must be in metaphase. They are black and white chromosomal karyotyping.

5. **There are *two types* of chromosomes**
A. *Autosomal* chromosomes: The word "soma" means body. They have influence on body characters. They are associated with structure and function of all cells not associated with sex. There are 44 autosomal chromosomes.
B. *Sex* chromosomes: They determine the sex. There are two sex chromosomes.

6. **Importance** of karyotyping
A. To study differences in chromosome size, shape and structure.
B. Determination of sex
C. Diagnose chromosomal disorder.
a. Mutations
b. Trisomy 21—Down's syndrome
c. Sex chromosome
D. Chromosomal problems
a. *Trisomy:* Extra copy of chromosome
b. *Monosomy:* Missing copy of chromosome (only 1 copy in zygote)
c. *Partial addition:* Extra part of a chromosome
d. *Partial deletion:* Missing part of a chromosome.
e. *Inversion:* Piece of chromosome breaks off, flips around and reattaches (changes order of gene)
f. *Translocation*: Piece of chromosome breaks of, reattaches to a different chromosome.

SN-14 What are genetic disorders?

Introduction: Mapping of chromosomes depending upon length and position of centromere is called karyotyping.

1. **Procedure:** It is done by the microculture of lymphocytes. The cells are grown in culture media *phytohaemagglutinin (PHA)*. The cell division is arrested in metaphase by adding colchicine. The spreads of the chromosome are counted and photographed. The images of each chromosomes are cut out and arranged as per classification.

   Karyotyping is done on the basis of

   A. Total length of chromosomes.

   B. Position of centromere.

   C. Relative length of two arms.

   D. Banding pattern.

   E. The chromosomes are arranged according to their length in a descending order. Identical chromosomes are paired in kcaryotyping. Then chromosomes paired are numbered 1 to 22 in descending order of length, i.e. pair No.1 is long, pair No. 22 is short. They are grouped into 7 groups. They are noted as A to G. The chromosomes are placed separately.

Table 9.5: Classes of chromosome

Group	Chromosome number	Features
A	1 to 3	Large, metacentric
B	4 and 5	Large, submetacentric
C	6 to 12	Medium sized, submetacentric
D	13 to 15	Medium sized, acrocentric with satellite
E	16 to 18	Short submetacentric
F	19 and 20	Short metacentric
G	21 and 22	Very short acrocentric

2. **Karyotyping helps to**

   A. Identify pattern of abnormal chromosome.

   B. Determination of the sex.