Subject: Biology

Grade: 12

Section: LS

Teacher: Abdallah Nassour

Unit: Reproduction and Genetics

Chapter 5: Human genetics

Document 3: Chromosomal Mutations

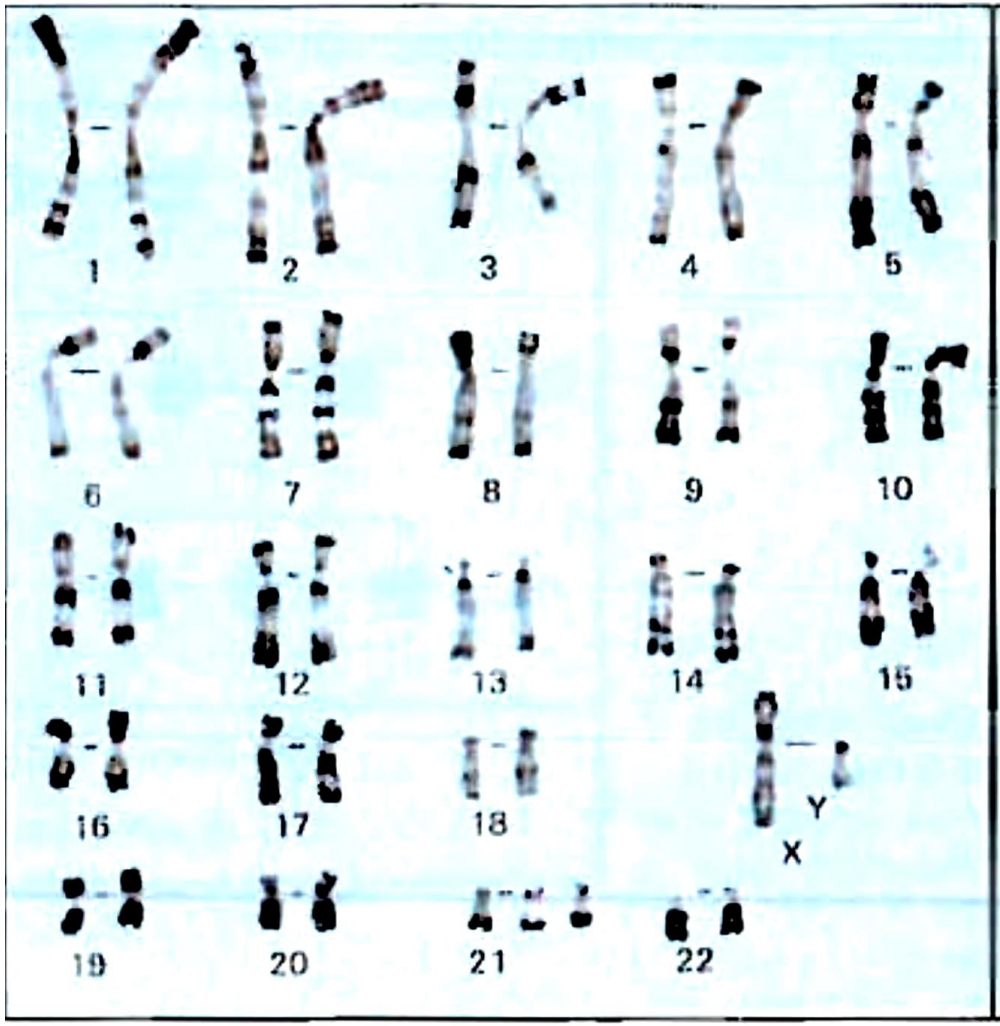
Gene mutations are a source of change in the genome. However, modifications of chromosomes in a cell, referred to as chromosomal mutations can also remodel the genome.

What are the different types of chromosomal mutations?

1– Abnormalities in chromosome number

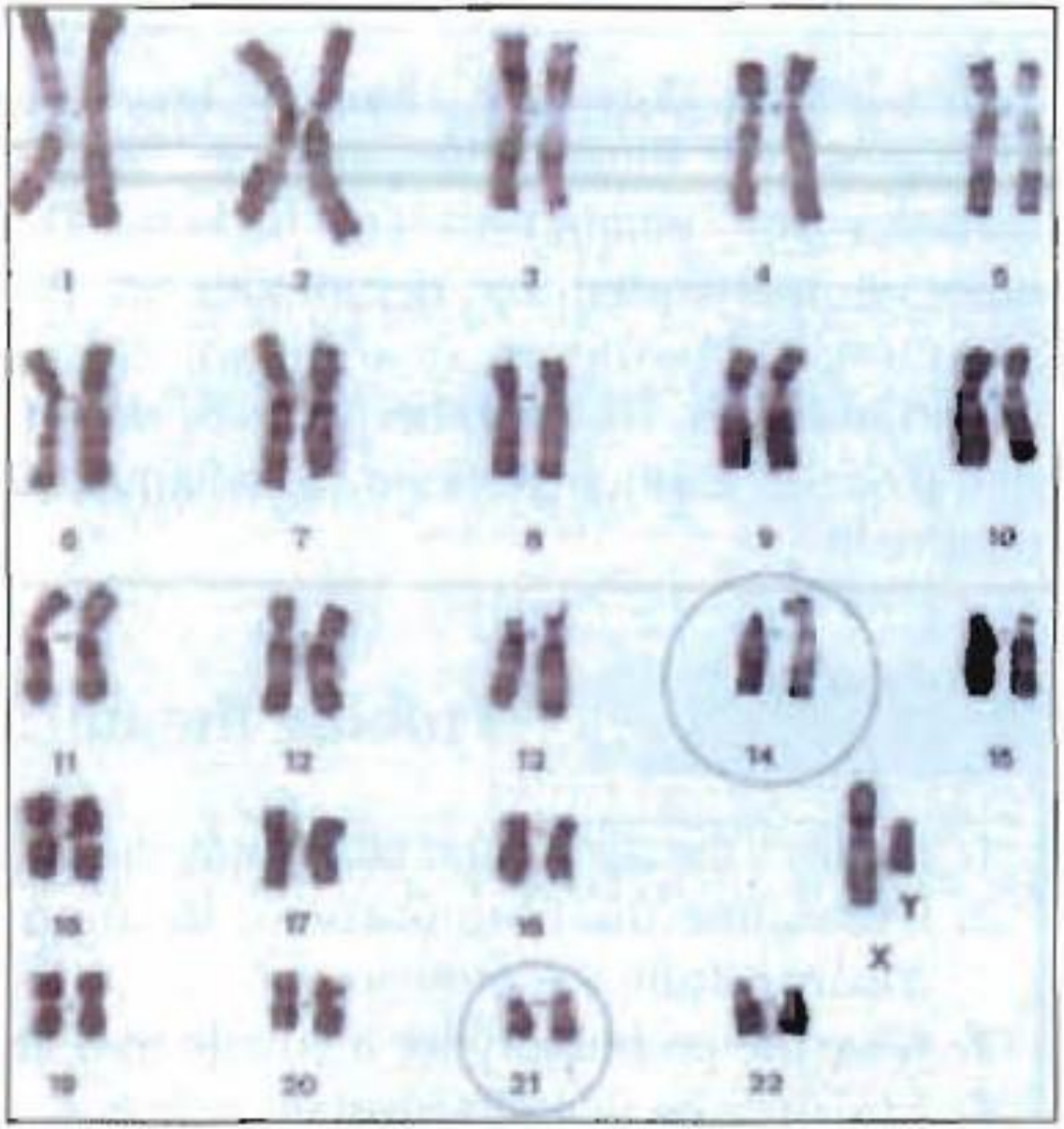
- **Autosomal abnormalities**

- Trisomy 21 is also called familial **Down syndrome** *(Doc.* a) after John Down, the British physician who described the condition in 1866. Affected individuals have 3 copies of chromosome 21. They show the following symptoms: oblique eyes, short hands with a unique palm crease, cardiac deformities and mental retardation. Sexual maturity is usually attained. The frequency of occurrence is 1 in 700 births but it varies depending on many factors *(Doc. b).*



Doc.a *Karyotype of a child with familial (free) trisomy 21.*

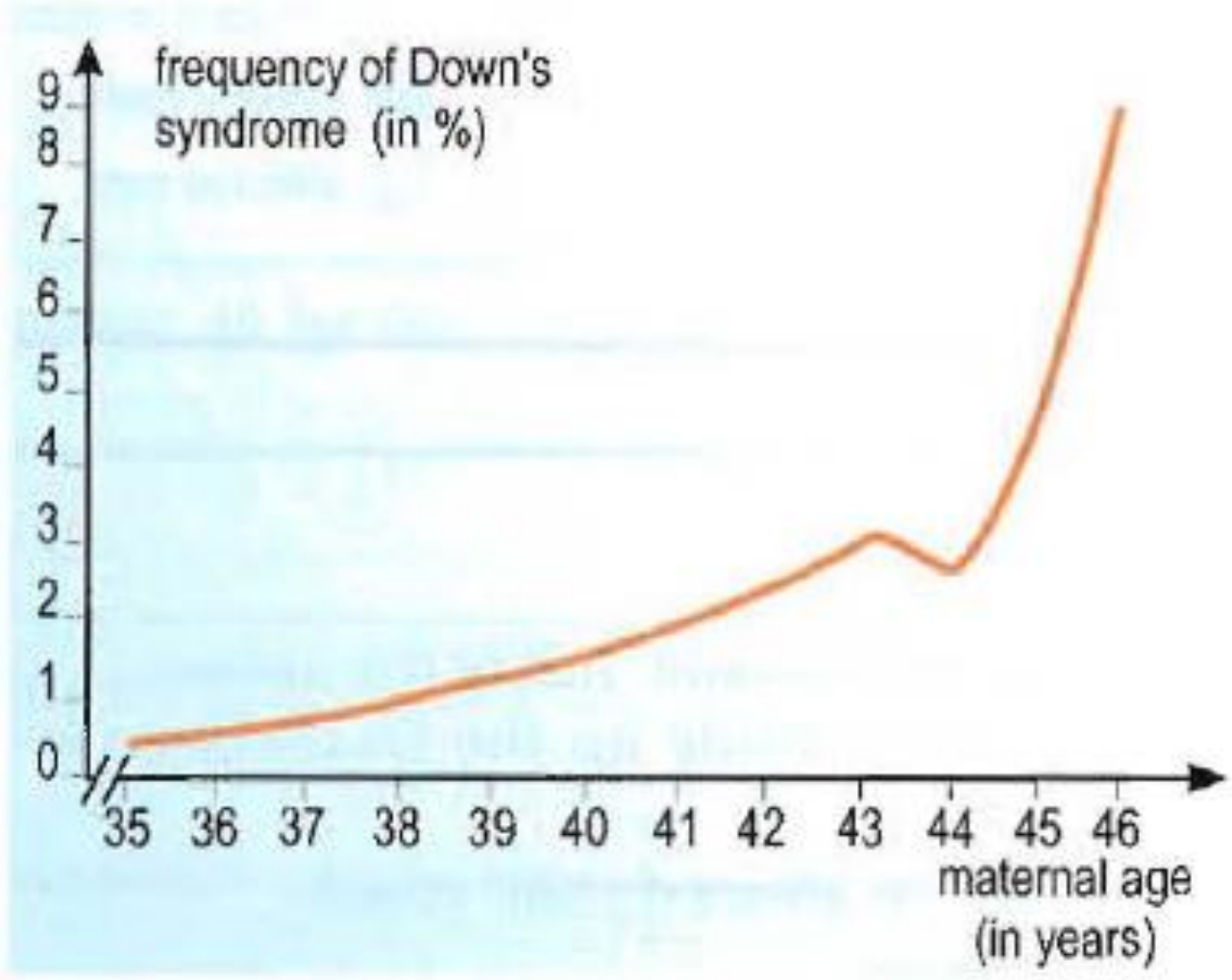
- Another type of trisomy 21 can result from **translocation** of chromosomes *(Doc.* c) Affected individuals have the same symptoms as in familial trisomy 21; however, their karyotype has in addition to the pair of chromosomes 21 , a third chromosome 21 attached (translocated) to another chromosome.



Doc.c *Karyotype of a child with linked (translocated) trisomy 21.*

What conclusion can we draw from *doc.b1*

the frequency of Down's syndrome as a function of maternal age increases from 0.5% at the age of 35 years to 3% at the age of 43. This increase is faster from this age onwards and reaches 8.5% at the age of 46. This shows that the frequency of this disease is enhanced by maternal age.



Doc.b *Variation of the frequency of trisomy 21 as a*

Therefore, the maternal age affects the frequency of Down's syndrome

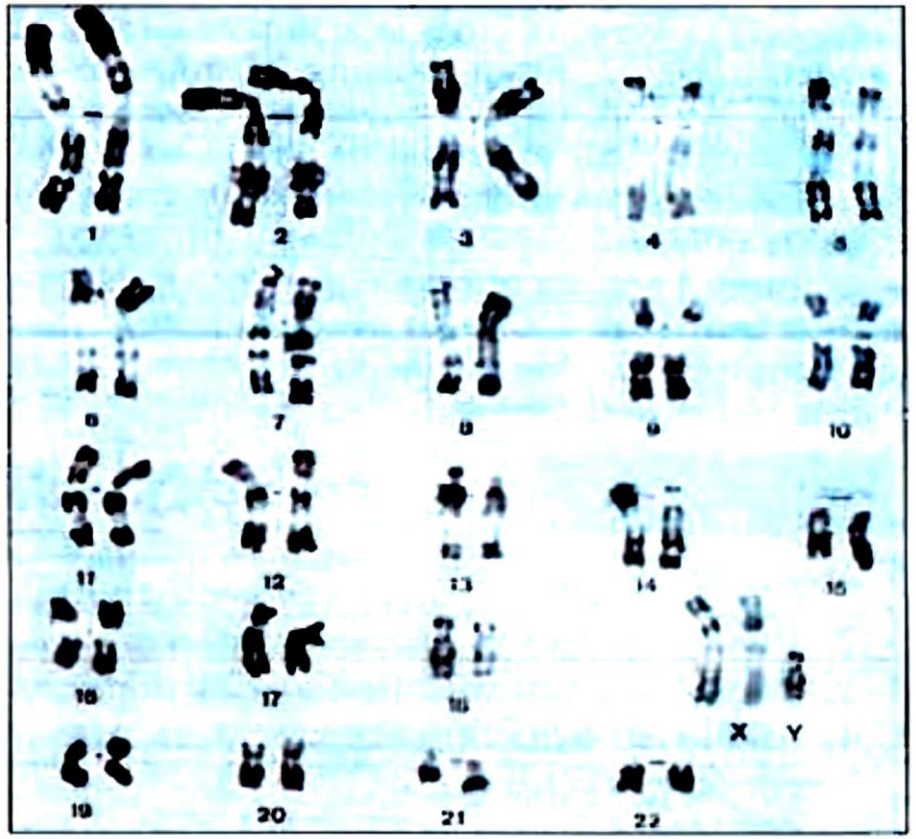
Sex chromosome gonosome abnormalities

Other abnormalities in chromosome number may affect the sex chromosomes, resulting in several syndromes:

Doc.e Karyotype of a woman having Turner syndrome.

-**Turner syndrome** (X 0) affects women, Patients have a short stature and are sterile due to the atrophy of the ova ries, Secondary sexual characteristics do not develop,

Other abnormalities are known, affecting the number of sex chromosomes such as XYY, XXX, etc.

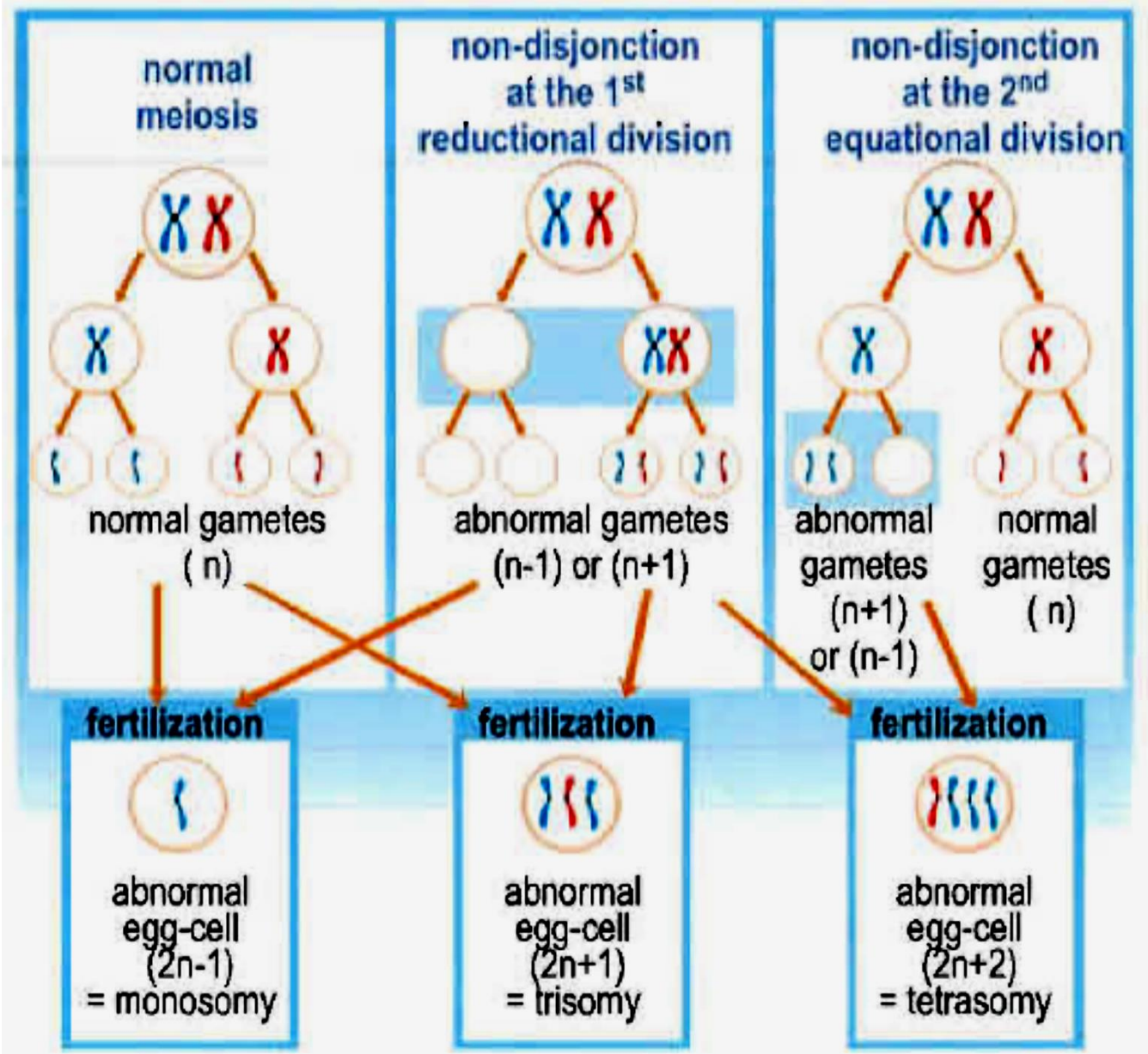


Doc.f Karyotype of a male having Klinefelter syndrome.

Klinefelter syndrome (XXY) affects males, who are sterile due to the atrophy of the testes. They exhibit both male and female secondary sexual characteristics (abnormal development of breast) and often mental retardation

Causes of abnormalities in chromosome number

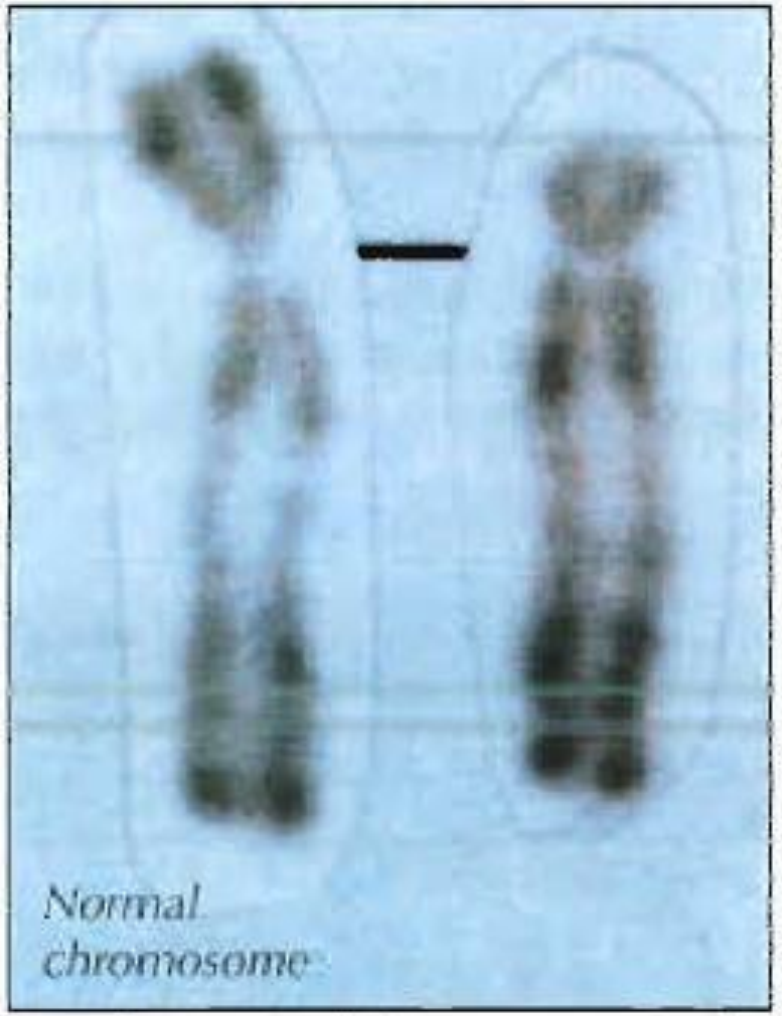
Abnormalities in chromosome number are usually due to errors in meiosis during gametogenesis. A non-disjunction of the homologous chromosomes leads to the formation of gametes that carry both copies of a chromosome and gametes that lack a copy of this chromosome. The union of these gametes with a normal gamete during fertilization produces zygotes that have respectively three copies of the same chromosome (trisomy) or a single copy of the chromosome (monosomy).



Doc.g Mechanisms of occurrence of chromosomal numerical abnormalities.

1– Abnormalities affecting the structure of chromosomes

The "cri-du-chat" disease (cat cry syndrome) causes severe mental retardation and larynx malformations so that a baby emits sounds like a cat's mewing. The karyotype of affected individuals shows an abnormality of one of the copies of human chromosomes 5.



Doc.b *A pair of chromosomes 5 of a child affected with the "cri- du-chat" (cat cry) syndrome.*

1. Compare the karyoypes of *doc.a* and *c*

- The karyotype shown in the doc, a presents an abnormality in the number of chromosomes. The chromosomal formula is 47 chromosomes and not 46 chromosomes. This is the karyotype of a male with extra chromosome 21. There are three copies of chromosome 21 and not two. This is the karyotype of an individual affected by trisomy 21 or Down syndrome (free trisomy 21).
- The karyotype in doc. C shows a structural abnormality of chromosomes. The chromosomal formula is 46 (2n = 46). All chromosomes are found in two copies, and are classified into homologous pairs except for chromosome 14 where one of the chromosome is longer than the other .An extra chromosome 21 is attached to Chromosome 14. this is a translocation 1421.(trisomy by translocation)
- In both cases, the amount of genetic material (total number of genes) is higher than the normal one: an extra chromosome 21 is present.

3- Write the chromosome formula (karyotype) of the syndromes represented in *doc.e* and f.

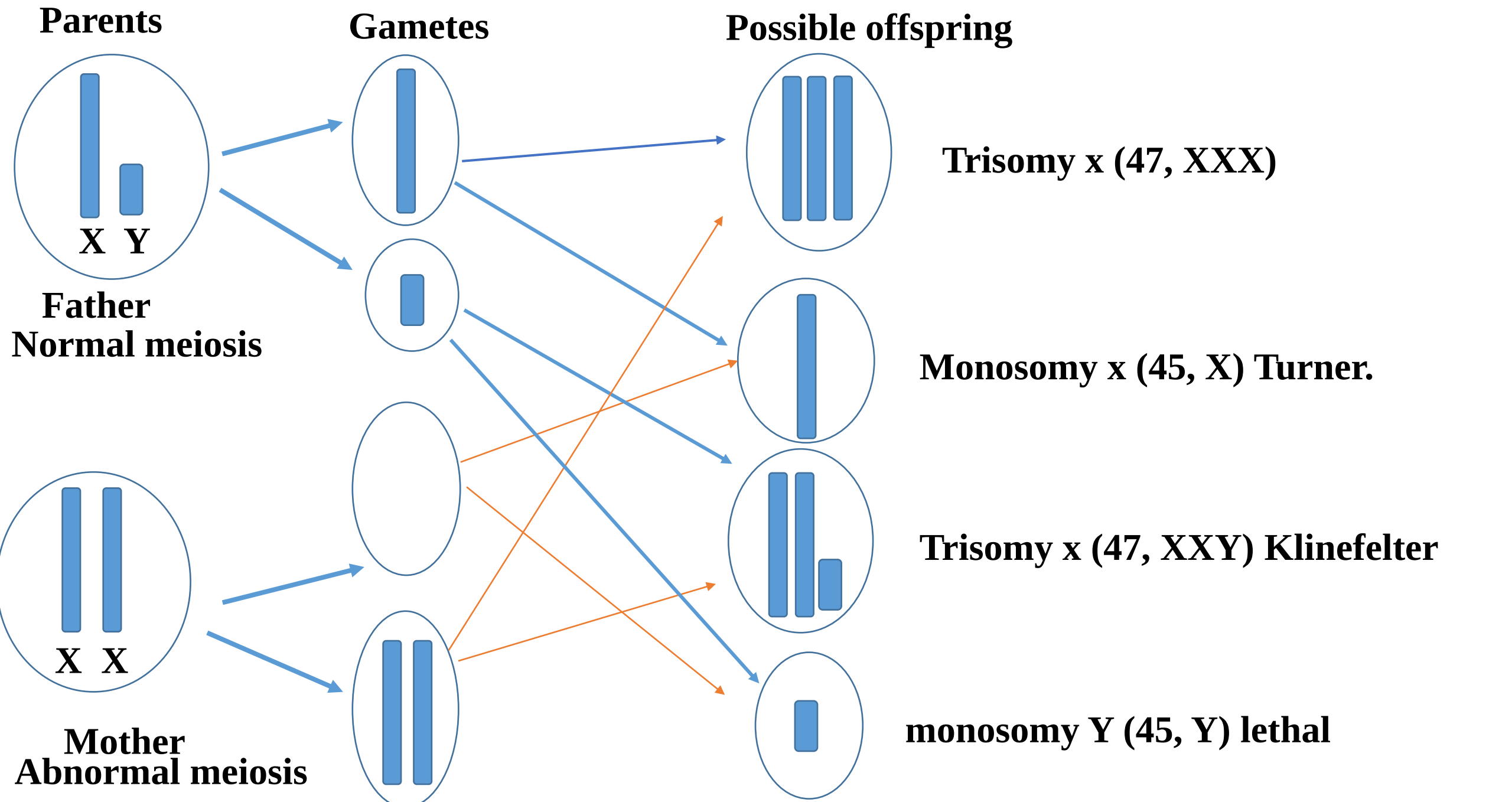
Trisomy 21= 47, XY, +21

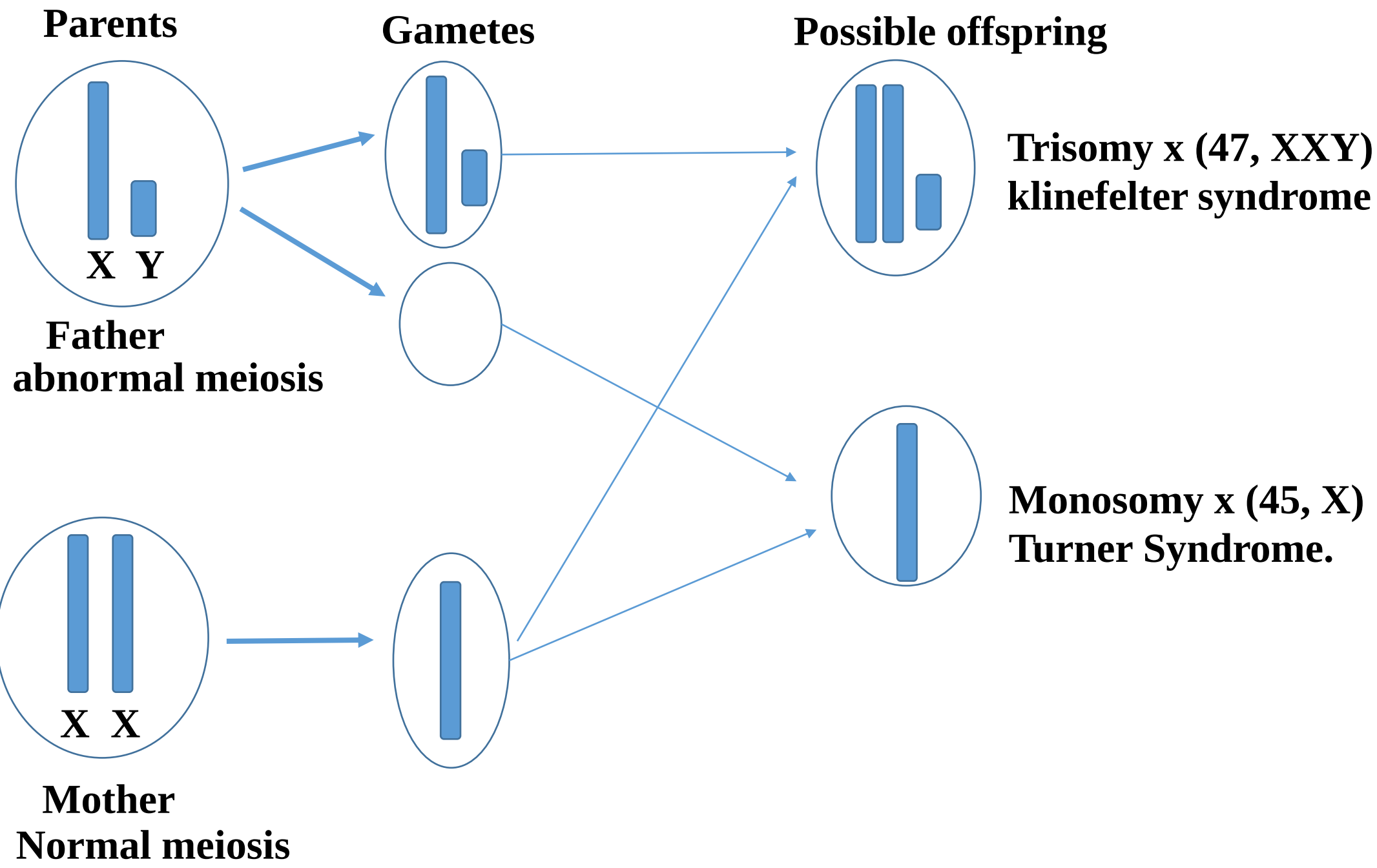
Monosomy 15 = 45, XX, -15

Turner syndrome (monosomy X) = 45, X

Klinefelter syndrome (trisomy) = 47, XXY

4- Basing on *doc.g*, illustrate in a diagram meiosis and fertilization processes which led to a klinefelter syndrome




Parents
Gametes
Possible offspring
X Y
Father
abnormal meiosis
Trisomy x (47, XXY)
klinefelter syndrome
Monosomy x (45, X)
Turner Syndrome.
X X
Mother
Normal meiosis

5- What is the type of mutation observed in *doc. h.* What is the percentage of abnormal gametes produced by this individual

One of the pair of chromosomes 5 ha lost most of irs short arm: this loss is called a “deletion” and is responsible fro : cri – du – chat” syndrome.
The percentage of abnormal gametes produced by this individual, which carry the abnormal chromosome responsible for the diseased is 50 %