Zoo-342 Molecular biology

Lecture 5

Changes in chromosome structure and number

Variation in chromosome structure: an overview

- Changes in chromosome structure and number are called **chromosomal aberration** or **chromosomal mutation**.
- Chromosomes may break due to:

1) ionizing radiation (physical agent) or
2) chemical compounds.

- Every chromatid break produces two ends. These ends have been described as **sticky**.

If broken chromatids ends are not brought together the following ways could occur:

1) They will remain broken, which will result in the loss of some of the genetic materials, which will affect the phenotype of the cell or organism.
2) They may rejoin in several different ways:
    a) The two broken ends of a single chromatid can reunite.
    b) The two broken end of one chromatid can fuse with the broken end of another chromatid, resulting in an exchange of a chromosomal material.

Types of breaks

I. Noncentromeric breaks (include):

A. Single breaks

1. Restitution
2. Deletion

B. Two breaks in the same chromosome

1. Deletion
2. Inversion

C. Two breaks in nonhomologous chromosomes

1. Reciprocal translocation

II. Centromeric breaks

A. Robertsonian translocation or fusion

Single breaks:

- If a single chromosome breaks, the broken chromosome is simply repaired (rejoin) in a process called **restitution.**
- When the broken ends do not rejoin, the result is an **acentric fragment**, **without** a **centromere** and a **centric fragment**, **with a centromere** (Figure 1).
- Missing of a part of chromosome is termed a **deletion**.

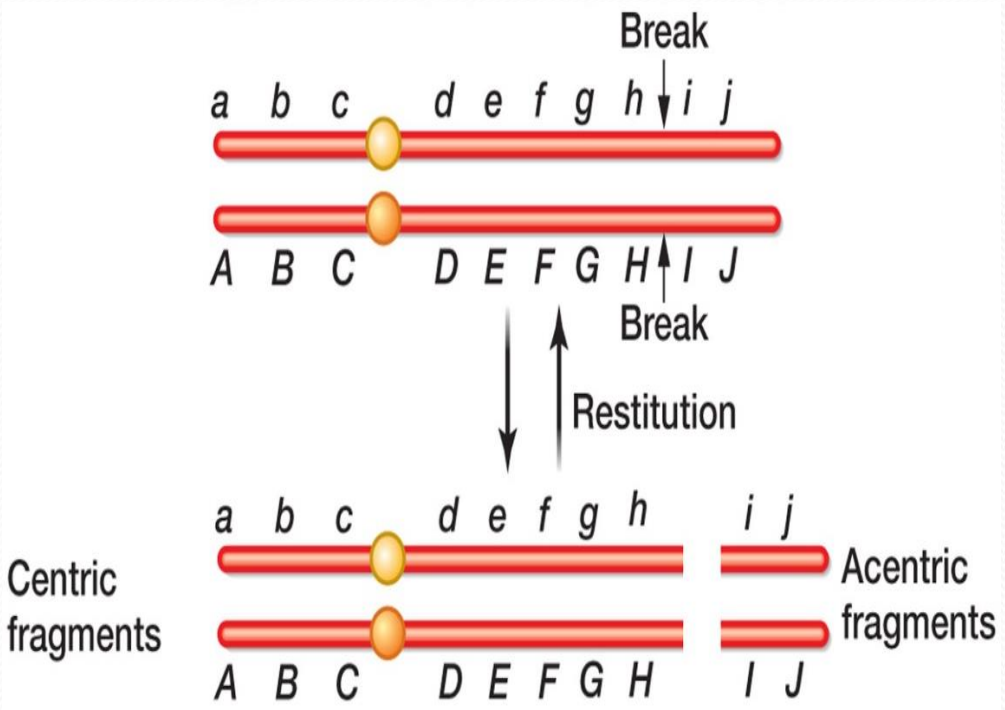

Figure 1: The consequences of a chromosome break in homologous chromosomes

Two breaks in the same chromosome:

Three possible consequences of a double break in the same chromosome (Figure 2):

1)**Inversion**: a chromosome breaks in two places and the broken fragment rejoins the same chromosome, but upside down (reversed). This is called a chromosome **inversion**.

2)**Deletion:** loss of segment of chromosome. The syndrome known as **cri du chat** (from the French, meaning cry of the cat) is so named because of the cat-like cry. This syndrome arises from a deletion in the short arm of chromosome 5; the karyotype for this syndrome is 46,XX or XY, $5p^-$ (Figure 3).

3)**Restitution**.

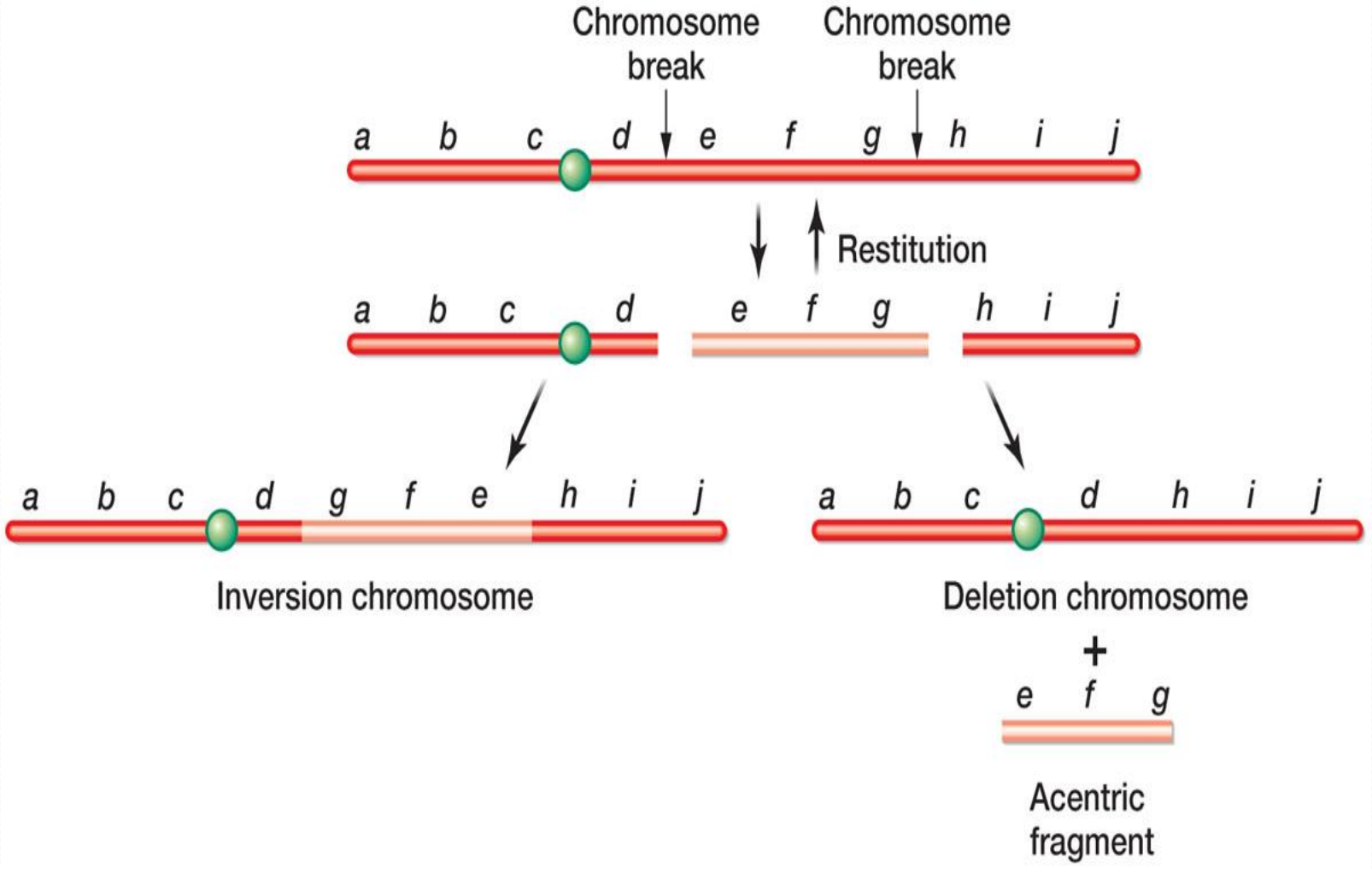

Figure 2: Three possible consequences of a double break in the same chromosome

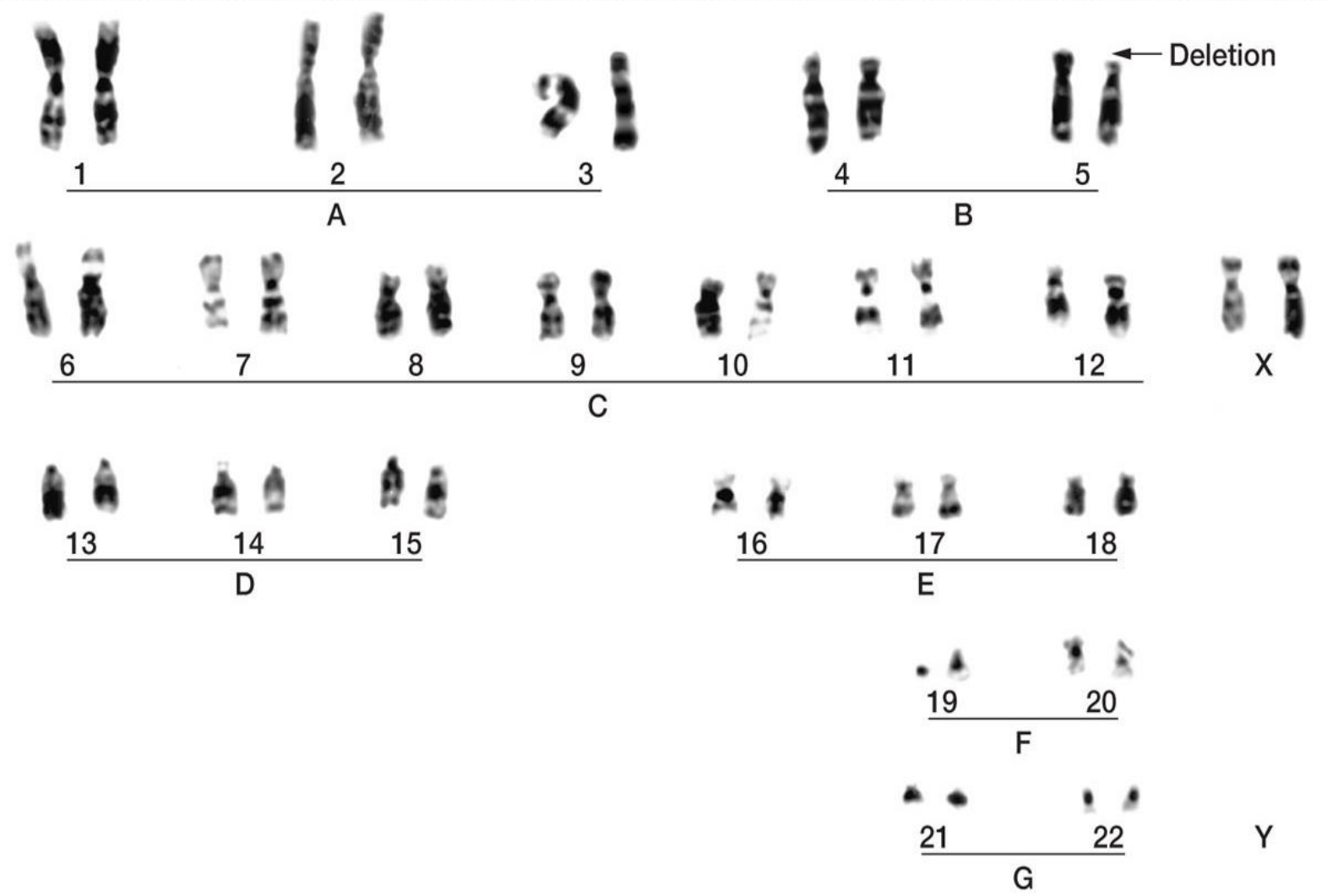

Figure 3: Karyotype of a girl with cri du chat syndrome

Two breaks in nonhomologous chromosome:

- Reciprocal translocations are usually an exchange of material between nonhomologous chromosomes (Figure 4).

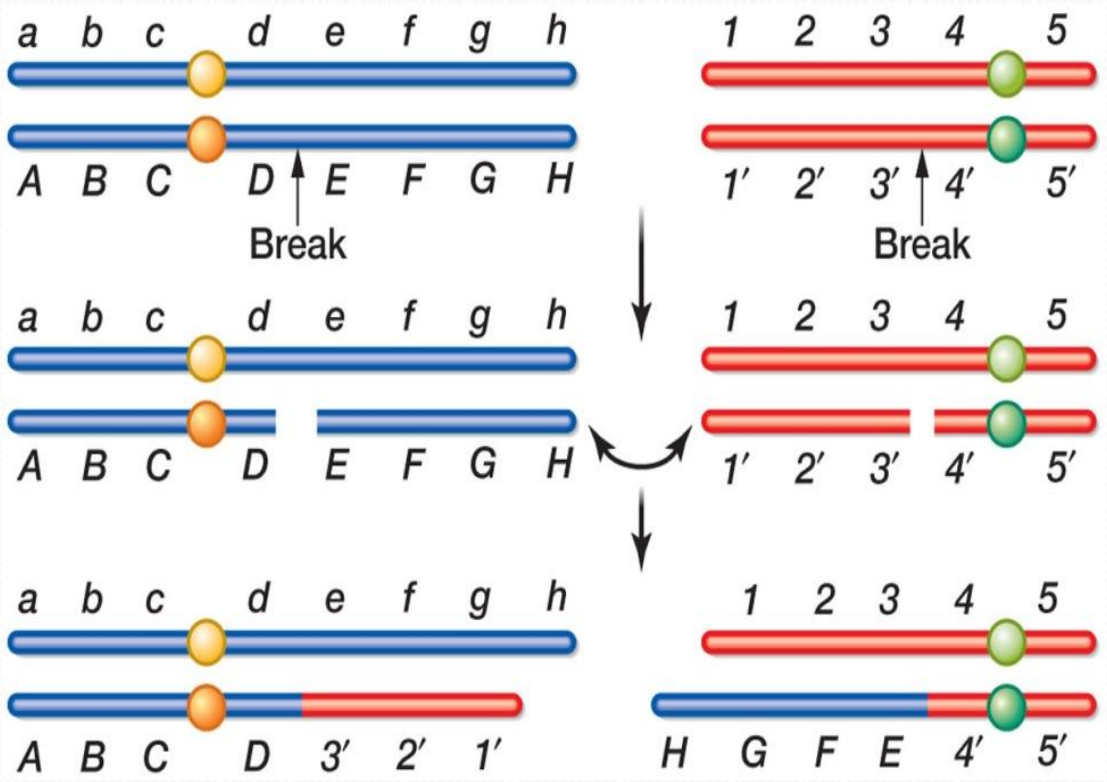

Figure 4: Generation of a reciprocal translocation

Centromeric breaks:

- **Robertsonian translocation** or **fusion** is a type of translocation caused by breaks at or near the centromeres of two **acrocentric** chromosomes.
- The **long** chromosomal arms from two acrocentric chromosomes are joined together, while the **short** chromosomal arms are lost because they both lack a centromere.
- **Robertsonian translocation** produces a decreased number of chromosomes because the long arms of two chromosomes are fused into one.
- In humans, Robertsonian translocation can occur between any two of the acrocentric chromosomes 13, 14, 15, 21, and 22.

Variation in chromosome number: an overview:

- Abnormalities of chromosome number occur as either **aneuploidy** or **euploidy**.
- **Aneuploidy** involves changes in chromosome number by additions or deletions of one or more individual chromosome.
- **Euploidy** involves changes in the number of sets of chromosomes.
- **Aneuploidy** results from nondisjunction during meiosis (Figure 5).

Types of aneuploidy:

1)**Monosomic**: diploid cell missing a single chromosome (2n-1).

2)**Nullisomic**: diploid cell missing two copies of chromosome (homologous chromosome) (2n-2).

3)**Trisomic**: diploid cell with an extra chromosome (2n+1).

4)**Tetrasomic**: diploid cell with two extra chromosomes (2n+2).

Euploidy:

Organisms with higher numbers of sets, such as triploids (3n) and tetraploids (4n), are called **polyploidy**

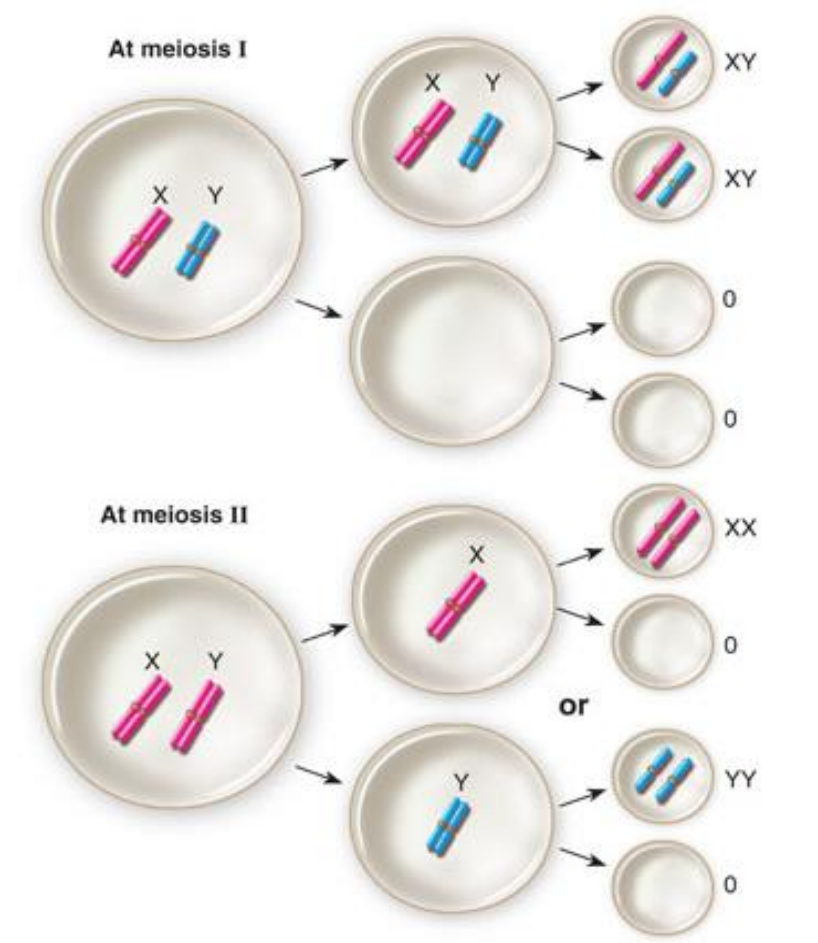

Figure 5: Nondisjunction of the sex chromosome in humans male

Changes in aneuploidy:

- Approximately 50% of the spontaneous abortion (miscarriages) among women involve fetuses with some chromosomal aberration, about half of these are autosomal trisomics.
- There are five types of human syndromes:
  1) Trisomy 21 (Down syndrome) (47, XX or XY, +21), children with Down syndrome have three copies of chromosome 21 (Figure 6).
  2) Trisomy 18 (Edward syndrome): The karyotype for this syndrome is 47, XX or XY, +18.
  3) Trisomy 13 (Patau syndrome): The karyotype for this syndrome is 47, XX or XY, +13.
  4) XO (Turner syndrome): The karyotype for this syndrome is 45, X (Figure 7).
  5) XXY (Klinefelter syndrome): The karyotype for this syndrome is 47, XXY.

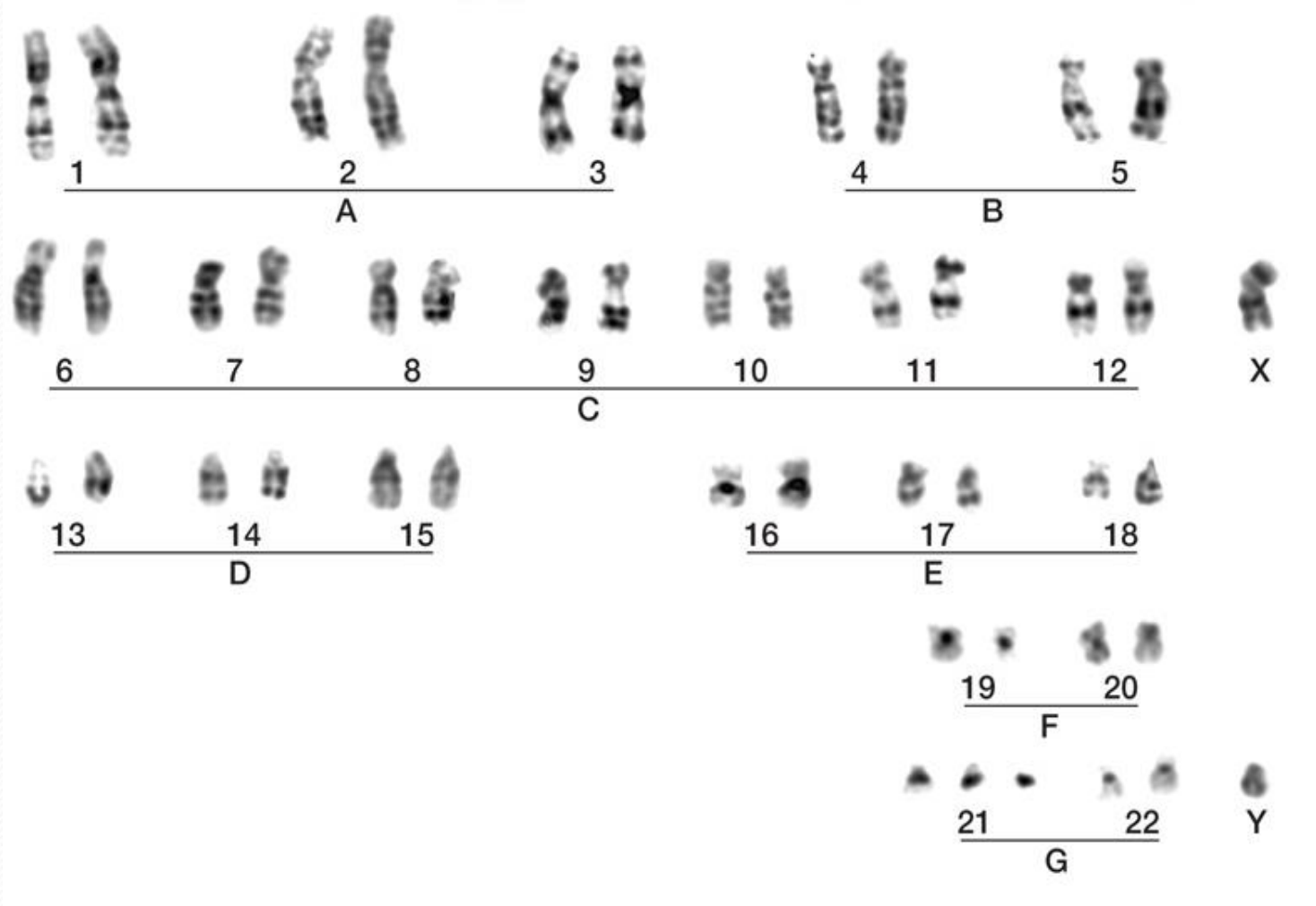

Figure 6: Karyotype of a male with Down syndrome

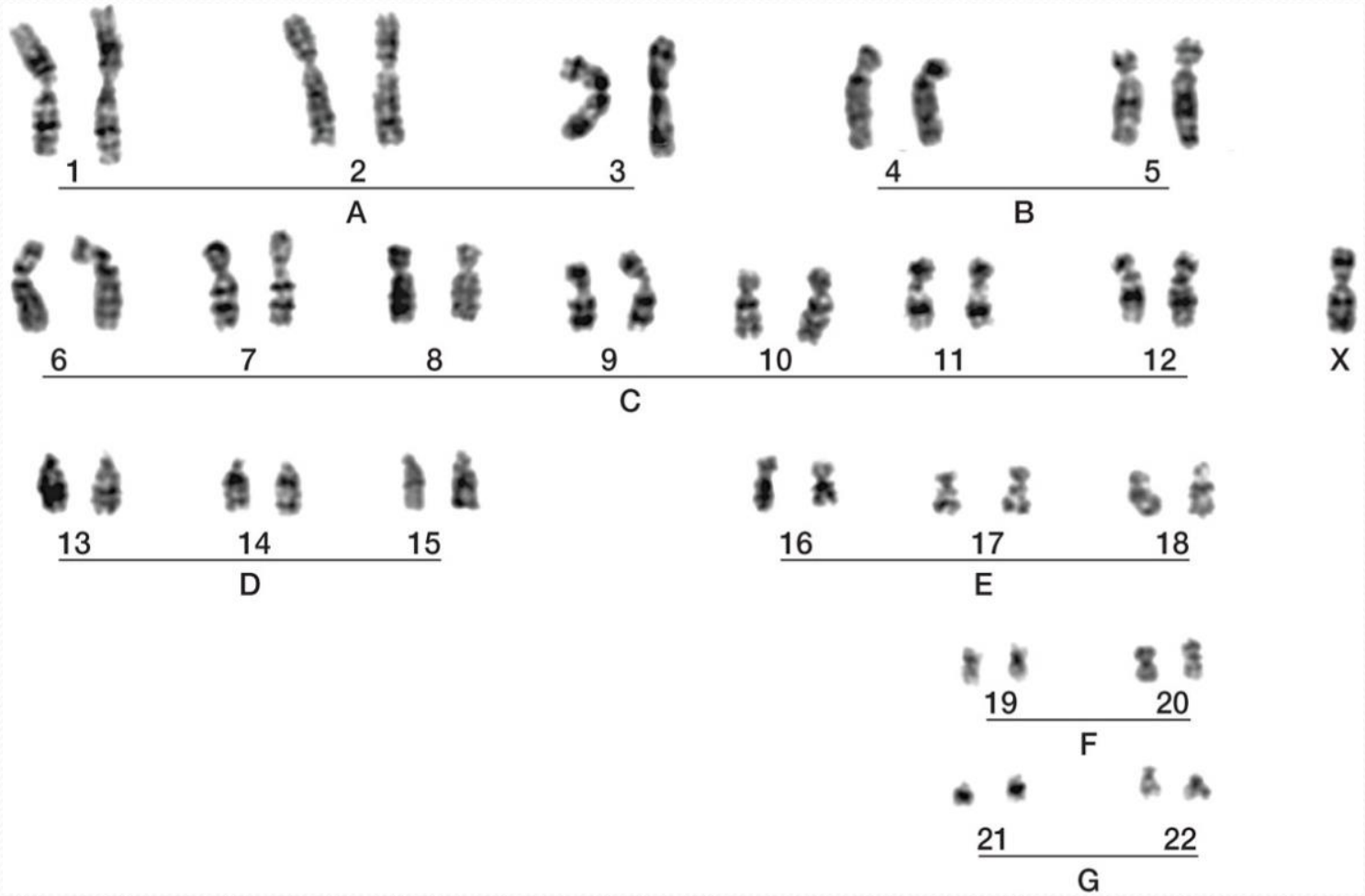

Figure 7: Karyotype of a female with Turner syndrome