# Human karyotype

# A karyotipe and its properties

- A **karyotype** is the number and appearance of chromosomes in the nucleus of a eukaryotic cell. The term is also used for the complete set of chromosomes in a species or in an individual organism and for a test that detects this complement or measures the number.
- Karyotypes describe the chromosome count of an organism and what these chromosomes look like under a light microscope. Attention is paid to their length, the position of the centromeres, banding pattern, any differences between the sex chromosomes, and any other physical characteristics. The preparation and study of karyotypes is part of cytogenetics.

# The karyotype

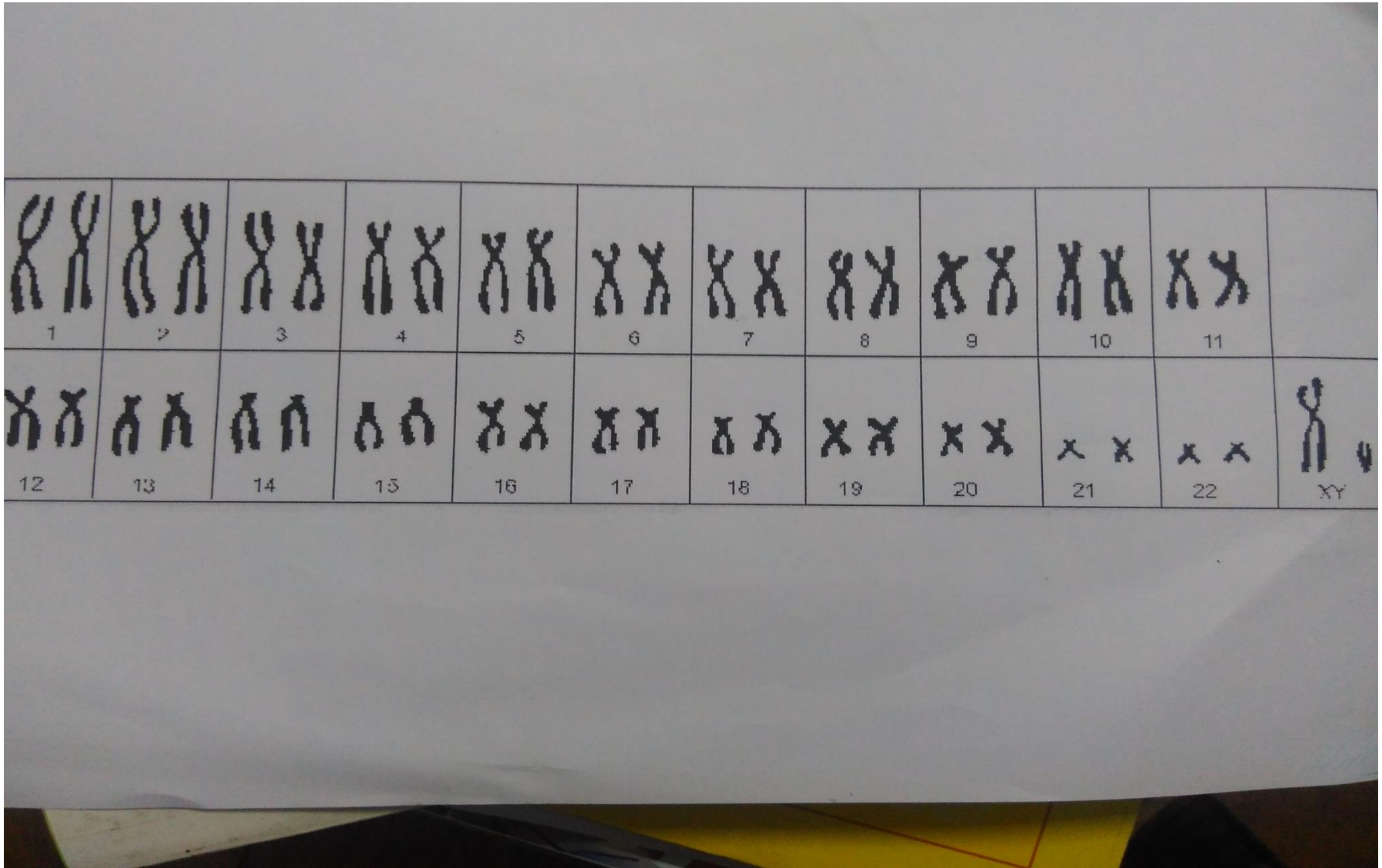
- The study of karyotypes is important for cell biology and genetics, and the results may be used in evolutionary biology and medicine. Karyotypes can be used for many purposes; such as to study chromosomal aberrations,

  cellular function, taxonomic relationships, and to gather information about
  past evolutionary events.

# Human karyotype

- The normal human karyotypes contain 22 pairs of autosomal chromosomes and one pair of sex chromosomes (allosomes). Normal karyotypes for females contain two X chromosomes and are denoted 46,XX; males have both an X and a Y chromosome denoted 46,XY. Any variation from the standard karyotype may lead to developmental abnormalities.

# Karyogram of human male using Giemsa (Geman chemist) staining



# Ideograms

- **An ideogram is a diagrammatic representation** of the karyotype that shows all of the pairs of homologous chromosomes in the nucleus. The pairs of chromosomes are lined up in order of size, so that the centromeres are aligned and the short arm is uppermost. An ideogram is a useful point of reference for analyzing mutations.

# Sex inheritance

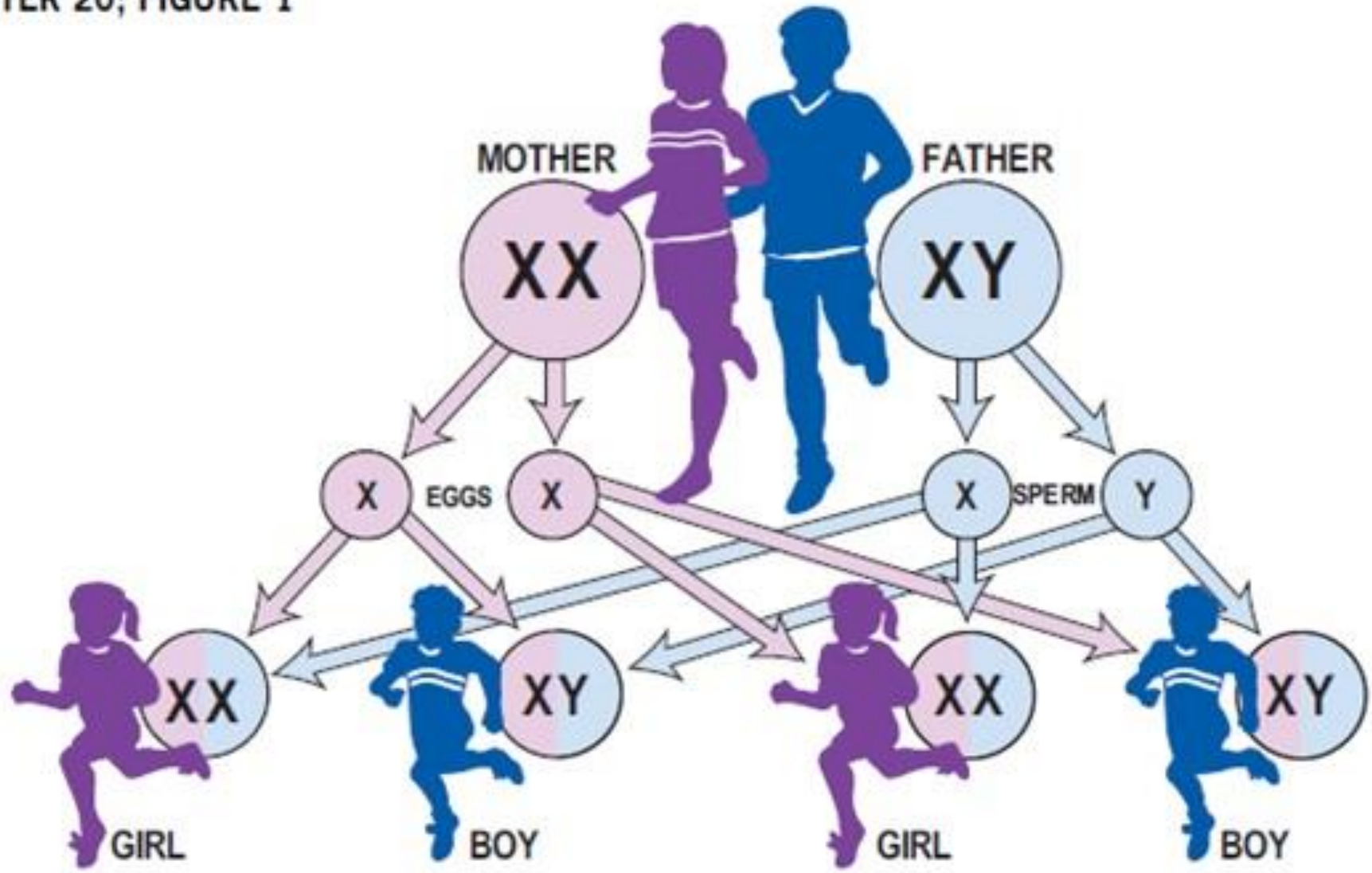
- In humans, fruit flies, XX = female; XY = male.
- Inheritance of sex is just like any other trait, except it involve inheritance of an entire chromosome.

Because there are genes on sex chromosomes, inheritance of certain traits can be sex-linked.

| | | Male parent: X | Male parent: Y |
|---|---|---|---|
| Female parent | X | XX | XY |
| | X | XX | XY |

# The Sex Chromosomes

CHAPTER 20; FIGURE 1



# Sex linkage

- **Sex linkage** is the phenotypic expression of an allele (an **allele** is a variant form of a given gene) related to the allosome (sex chromosome) of the individual. In autosomal chromosomes both sexes have the same probability of existing, but since humans have many more genes on the female X chromosome than on the male Y chromosome, these are much more common than Y-linked traits.
- In mammals, the female is homogametic, with two X chromosomes (XX), while the male is the heterogametic sex, with one X and one Y chromosome (XY). Genes on the X or Y chromosome are called sex-linked.

- Linkage and recombination are phenomena that describe the inheritance of genes. A linkage is a phenomenon where two or more linked genes are always inherited together in the same combination for more than two generations. The recombination frequency of the test cross progeny is always lower than 50%. Therefore, if any two genes are completely linked, their recombination frequency is almost 0%. The phenomenon of linkage was studied by the scientist T.H. Morgan using the common fruit fly or *Drosophila melanogaster.*