In diploid organisms, somatic cells (non-sex-cells), have pairs of homologous chromosomes. Homologous chromosomes share shape and genetic loci, and carry genes that carry the same genetic characteristics.

- Each of the homologues is inherited from a separate parent.

**Note:** The sets are combined in the first cells following fertilization and passed down together from cell to cell during growth and development by mitosis.

- In humans, 22 pairs, found in males and females, are autosomes. Two other chromosomes are sex chromosomes.

- In mammalian females, there are two X chromosomes; in male mammals an X and a Y.
Gametes have a single set of chromosomes

- Adult animals have somatic cells with two sets of homologues (diploid, 2n).
- Sex cells (gametes = eggs & sperm) have one set of homologues (haploid, n). These cells are produced by meiosis.
- Sexual lifecycles involve the alteration between a diploid phase and haploid phase.
- The fusion of haploid gametes in the process of fertilization results in the formation of a diploid zygote.

Meiosis reduces the chromosome number from diploid to haploid

- An understanding of the cell cycle is needed for an understanding of meiosis.
- Meiosis occurs only in diploid cells.
- Like mitosis, meiosis is preceded by a single duplication of the chromosomes.
- The overall result is four daughter cells, each with half the number of chromosomes.

Again, the process is dynamic but may stop at certain phases for long periods of time.

- The process includes two consecutive divisions (meiosis I and meiosis II).
- The halving of the chromosome number occurs in meiosis I. The end result is two haploid cells, with each chromosome consisting of two chromatids.
- Sister chromatids separate in meiosis II.
- The end result is four haploid cells.

A comparison of mitosis and meiosis

- All the events unique to meiosis occur in meiosis I. In prophase I, homologous chromosomes pair to form a tetrad, and crossing over occurs between the homologous chromatids.

  **Note:** This results in unique genetic combinations.

- Meiosis II is virtually identical to mitosis (except cells are haploid).
• Mitosis results in two daughter cells, each with the same chromosomes as the parent cell. Mitosis can occur in haploid or diploid cells.

• Meiosis results in four daughter cells, each with half the number of chromosomes as the parent cell. Meiosis occurs only in diploid cells.

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Independent orientation of chromosomes in meiosis and random fertilization lead to varied offspring

• During prophase I of meiosis, each homologue pairs up with its “other.” During this process, X and Y chromosomes behave as a homologous pair.

  **Note:** This pairing of homologues is called synapsis.

• When they separate at anaphase I, maternally and paternally inherited homologues move to one pole or the other independently of the other pairs.

• Given n chromosomes, there are 2^n ways that different combinations of the half-pairs can move to one pole.

• In humans, there are 2^{23} ways of combining an individual’s maternally inherited and paternally inherited homologues.

• Combining gametes into zygotes suggest there are 2^{23} x 2^{23} combinations in the zygote.

Homologous chromosomes carry different versions of genes

• Simplified examples: coat color and eye color in mice.

  • C (brown) and c (white) for different versions of the coat-color gene and E (black) and e (pink) for different eye color genes.

• In this example, with the information up to this point, there would be two possible outcomes for the genes on the two chromosomes in a gamete (C'E' and Ce).
Crossing over further increases genetic variability

• Crossing over is the exchange of corresponding segments between two homologues (sister chromatid exchange). The site of crossing over is called a chiasma.
• This happens between chromatids within tetrads as homologues pair up during synapsis (prophase I).
• Crossing over produces new combinations of genes (genetic recombination).

• Because crossing over can result in variable locations among thousands of genes in each tetrad, variability is great. Essentially, two individual parents could never produce identical offspring from two separate fertilizations.

Note: It is for this reason that, with exception of identical twins everyone is a unique genetic entity never seen before and never to be seen again.

A karyotype is a photographic inventory of an individual’s chromosomes

• Blood samples are cultured for several days under conditions that promote cell division of white blood cells.
  
  NOTE: Red blood cells lack nuclei and do not divide.

• The culture is treated with a chemical that stops cell division at metaphase.

• White blood cells are separated, stained, and squashed (to spread out the chromosomes) following the procedure.

• From this the genetic sex of an individual can be determined and abnormalities in chromosomal structure and number can be detected.

An extra copy of chromosome 21 causes Down syndrome

• In most cases human offspring that develop from zygotes with an incorrect number of chromosomes abort spontaneously.
• Trisomy 21 is the most common chromosome-number abnormality, occurring in about one out of 700 births.

• Down syndrome includes a number and range of physical, mental, and disease susceptibility features.

• The incidence of Down syndrome increases with the age of the mother.
Accidents during meiosis can alter chromosome number

- **Nondisjunction** is the failure of chromosome pairs to separate during either meiosis I or meiosis II.

- Fertilization of an egg resulting from nondisjunction with a normal sperm results in a zygote with an abnormal chromosome number.

Abnormal numbers of sex chromosomes do not usually affect survival

- Unusual numbers of sex chromosomes upset the genetic balance less than do unusual numbers of autosomes, perhaps because the Y chromosome carries fewer genes and extra X chromosomes are inactivated as Barr bodies in females.

- Abnormalities in sex chromosome number result in individuals with a variety of different characteristics, some more seriously affecting fertility or intelligence than others.

The greater number of sex chromosome abnormalities illustrates the crucial role of the Y chromosome in determining a person’s sex. A single Y is enough to produce “maleness” even in combination with a number of Xs, whereas the lack of Y results in “femaleness.”

The explanation for the increased incidence of trisomy 21 among older women is not entirely clear but probably involves the length of time a woman’s developing eggs are in meiosis. Meiosis begins in all eggs before the woman is born, and finishes as each egg matures in the monthly cycle following puberty. Eggs of older women have been “within” meiosis longer.
Alteration of chromosomes structure can cause birth defects and cancer

- Deletions, duplications, and inversions occur within one chromosome.

- Inversions are less likely to produce harmful effects than deletions or duplications because all the chromosome’s genes are still present.

- Duplications, if they result in the duplication of an oncogene in somatic cells, they may increase the incidence of cancer.

- Translocation involves the transfer of a chromosome fragment between nonhomologous chromosomes.

- Translocations may or may not be harmful. One type of translocation results in Down Syndrome.

- Chromosomal changes in somatic cells may increase the risk of cancer.