13.1C: Identification of Chromosomes and Karyotypes

A karyotype depicts the number, size, and any abnormalities of the chromosomes in an organism.

Learning Objectives

- Describe a normal human karyotype and discuss the various abnormalities that can be detected using this technique

Key Points

- A normal human karyotype contains 23 pairs of chromosomes: 22 pairs of autosomes and 1 pair of sex chromosomes, generally arranged in order from largest to smallest.
- The short arm of a chromosome is referred to as the p arm, while the long arm is designated the q arm.
- To observe a karyotype, cells are collected from a blood or tissue sample and stimulated to begin dividing; the chromosomes are arrested in metaphase, preserved in a fixative and applied to a slide where they are stained with a dye to visualize the distinct banding patterns of each chromosome pair.
- A karyotype can be used to visualize abnormalities in the chromosomes, such as an incorrect number of chromosomes, deletions, insertions, or translocations of DNA.

Key Terms

- **autosome**: any chromosome other than sex chromosomes
- **karyotype**: the observed characteristics (number, type, shape etc) of the chromosomes of an individual or species
- **translocation**: a transfer of a chromosomal segment to a new position, especially on a nonhomologous
Identification of Chromosomes

The isolation and microscopic observation of chromosomes forms the basis of cytogenetics and is the primary method by which clinicians detect chromosomal abnormalities in humans. A karyotype is the number and appearance of chromosomes. To obtain a view of an individual’s karyotype, cytologists photograph the chromosomes and then cut and paste each chromosome into a chart, or karyogram, also known as an ideogram.

In a given species, chromosomes can be identified by their number, size, centromere position, and banding pattern. In a human karyotype, autosomes or “body chromosomes” (all of the non-sex chromosomes) are generally organized in approximate order of size from largest (chromosome 1) to smallest (chromosome 22). However, chromosome 21 is actually shorter than chromosome 22. This was discovered after the naming of Down syndrome as trisomy 21, reflecting how this disease results from possessing one extra chromosome 21 (three total). Not wanting to change the name of this important disease, chromosome 21 retained its numbering, despite describing the shortest set of chromosomes. The X and Y chromosomes are not autosomes and are referred to as the sex chromosomes.

The chromosome “arms” projecting from either end of the centromere may be designated as short or long, depending on their relative lengths. The short arm is abbreviated p (for “petite”), whereas the long arm is abbreviated q (because it follows “p” alphabetically). Each arm is further subdivided and denoted by a number. Using this naming system, locations on chromosomes can be described consistently in the scientific literature.

Although Mendel is referred to as the “father of modern genetics,” he performed his experiments with none of the tools that the geneticists of today routinely employ. One such powerful cytological technique is karyotyping, a method in which traits characterized by chromosomal abnormalities can be identified from a single cell. To observe an individual’s karyotype, a person’s cells (like white blood cells) are first collected from a blood sample or other tissue. In the laboratory, the isolated cells are stimulated to begin actively dividing. A chemical called colchicine is then applied to cells to arrest condensed chromosomes in metaphase. Cells are then made to swell using a hypotonic solution so the chromosomes spread apart. Finally, the sample is preserved in a fixative and applied to a slide.

The geneticist then stains chromosomes with one of several dyes to better visualize the distinct and reproducible banding patterns of each chromosome pair. Following staining, the chromosomes are viewed using bright-field microscopy. A common stain choice is the Giemsa stain. Giemsa staining results in approximately 400–800 bands (of tightly coiled DNA and condensed proteins) arranged along all of the 23 chromosome pairs. An experienced geneticist can identify each chromosome based on its characteristic banding pattern. In addition to the banding patterns, chromosomes are further identified on the basis of size and centromere location. To obtain the classic depiction of the karyotype in which homologous pairs of chromosomes are aligned in numerical order from longest to shortest, the geneticist obtains a digital image, identifies each chromosome, and manually arranges the chromosomes into this pattern.
A human karyotype: This karyotype is of a male human. Notice that homologous chromosomes are the same size, and have the same centromere positions and banding patterns. A human female would have an XX chromosome pair instead of the XY pair shown.

At its most basic, the karyotype may reveal genetic abnormalities in which an individual has too many or too few chromosomes per cell. Examples of this are Down Syndrome, which is identified by a third copy of chromosome 21, and Turner Syndrome, which is characterized by the presence of only one X chromosome in women instead of the normal two. Geneticists can also identify large deletions or insertions of DNA. For instance, Jacobsen Syndrome, which involves distinctive facial features as well as heart and bleeding defects, is identified by a deletion on chromosome 11. Finally, the karyotype can pinpoint translocations, which occur when a segment of genetic material breaks from one chromosome and reattaches to another chromosome or to a different part of the same chromosome. Translocations are implicated in certain cancers, including chronic myelogenous leukemia.

During Mendel's lifetime, inheritance was an abstract concept that could only be inferred by performing crosses and observing the traits expressed by offspring. By observing a karyotype, today's geneticists can actually visualize the chromosomal composition of an individual to confirm or predict genetic abnormalities in offspring, even before birth.

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