Section Summary

A karyotype is the number and appearance of an individual's chromosomes, including their length, banding pattern, and centromere position. The number, size, shape, and banding pattern of chromosomes make them easily identifiable in a karyogram. A karyogram allows for the assessment of many chromosomal abnormalities. Disorders in chromosome number are typically lethal to the embryo, although a few trisomy and monosomy conditions are viable. Chromosome number abnormalities can occur because of nondisjunction, the failure of homologous chromosomes or sister chromatids to separate properly. Chromosomal structural abnormalities may also occur and include segments of the chromosome being duplicated, deleted, inverted, or translocated. All of these aberrations can result in problematic phenotypic effects.

Exercises

1. The genotype XXY would be:
    a. A monosomy condition
    b. A trisomy condition
    c. A deletion
    d. A polyploid

2. Nondisjunction is:
    a. failure of homologous chromosomes to separate properly
    b. is an example of a chromosomal rearrangement
    c. only occurs during meiosis II
    d. involves only autosomes

3. Polyploidy often happens in animal cells.
    a. True
    b. False

4. Explain what a karyotype is and why a karyogram helps identify different genetic conditions.

Answers

1. (b)
2. (a)
3. (b)
4. A karyotype is the number and appearance of an individual's chromosomes, including their length, banding pattern, and centromere position. Abnormalities in chromosome numbers are obvious when looking at a karyogram because it shows if an extra chromosome is present of whether an entire chromosome has been lost.

Glossary

aneuploid: an individual with an error in chromosome number; includes deletions and duplications of chromosome segments

deletion: a part of a chromosome is lost or removed

duplication: a part of a chromosome is duplicated and either inserted into a different position on the same chromosome or a completely different chromosome

euploid: an individual with the appropriate number of chromosomes for their species

inversion: the detachment, 180° rotation, and reinsertion of a chromosome arm

karyogram: the photographic image of a karyotype

karyotype: the number and appearance of an individual's chromosomes, including the size, banding patterns, and centromere position

monosomy: an otherwise diploid genotype in which one chromosome is missing

nondisjunction: the failure of synapsed homologs to completely separate and migrate to separate poles during the first cell division of meiosis

polyploid: an individual with an incorrect number of chromosome sets

translocation: the process by which one segment of a chromosome dissociates and reattaches to a different, nonhomologous chromosome

trisomy: an otherwise diploid genotype in which one entire chromosome is duplicated

Footnotes

1 V Goidts, et al., "Segmental duplication associated with the human-specific inversion of chromosome 18: a further example of the impact of segmental duplications on karyotype and genome evolution in primates," *Human Genetics*, 115 (2004):116–22.