

14.1 Human Chromosomes

Lesson Objectives

- Identify the types of human chromosomes in a karyotype.
- Describe the patterns of the inheritance of human traits.
- Explain how pedigrees are used to study human traits.

Lesson Summary

Karyotype: A genome is the full set of all the genetic information that an organism carries in its DNA. Chromosomes are bundles of DNA and protein found in the nucleus of a eukaryotic cell. A karyotype is a picture that shows the complete diploid set of human chromosomes, grouped in pairs and arranged in order of decreasing size. A typical human diploid cell contains 46 chromosomes, or 23 pairs:

- Two of the 46 are the **sex chromosomes** that determine an individual's sex: XX = female and XY = male. The X chromosome carries nearly 10 times the number of genes as the Y chromosome.
- The other 44 are **autosomes**, or **autosomal chromosomes**.

Transmission of Human Traits: Human genes follow the same Mendelian patterns of inheritance as the genes of other organisms:

- Many human traits follow a pattern of simple dominance.
- The alleles for many human genes display codominant inheritance.
- Many human genes, including the genes for blood group, have multiple alleles.
- A gene located on a sex chromosome is a **sex-linked gene**. The genes on sex chromosomes show a sex-linked pattern of inheritance, since females have two copies of many genes (located on X chromosomes) while males have just one.
- In females, most of the genes in one of the X chromosomes are inactivated in each cell.

Human Pedigrees: A chart used to analyze the pattern of inheritance that shows the relationships in a family is a **pedigree**. Pedigrees can be used to determine the nature of genes and alleles associated with inherited human traits.

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Eeva Therma, Millard Susman

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Human Chromosomes Orlando J. Miller, Eeva Therman, 2001 The fourth edition of this well known text provides students researchers and technicians in the area of medicine genetics and cell biology with a concise understandable introduction to the structure and behavior of human chromosomes. This new edition continues to cover both basic and up to date material on normal and defective chromosomes yet is particularly strengthened by the complete revision of the material on the molecular genetics of chromosomes and chromosomal defects. The mapping and molecular analysis of chromosomes is one of the most exciting and active areas of modern biomedical research and this book will be invaluable to scientists students technicians and physicians with an interest in the function and dysfunction of chromosomes. *Human Chromosomes* Audrey

Bishop, Patricia Cooke, 2014-05-12 *Human Chromosomes An Illustrated Introduction to Human Cytogenetics* focuses on the processes methodologies and approaches involved in the study of human chromosomes. The publication first offers information on the cell and its activity particularly noting that the cell is the basic unit that forms the organs and tissues of the human body. The differentiation of cells and the process of cell division are discussed. The text then focuses on the culture of human cells for the investigation of the chromosomes. The book elaborates on the identification of human chromosomes including further methods of identification and the use of radioactive isotopes. The publication also ponders on the numerical changes in the karyotype structural changes and X chromosomes. Discussions focus on the processes of mitosis and meiosis translocation deletion duplication and ring formation and the behavior transformation and characteristics of X chromosome. The text is a valuable reference for researchers interested in the study of human chromosomes. *Human Chromosomes* Eeva Therman, Millard Susman, 1993 Providing students researchers and technicians in the areas of medicine genetics and cell biology with a concise understandable introduction to the structure and behavior of human chromosomes this new edition has been extensively revised. It includes recent information in the field of chromosomal molecular genetics and will be invaluable to anyone with an interest in the function and dysfunction of chromosomes. 105 illustrations

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