

14.3 Studying the Human Genome

Lesson Objectives

- ☑ Summarize the methods of DNA analysis.
- ☑ State the goals of the Human Genome Project and explain what we have learned so far.

Lesson Summary

Manipulating DNA Since the 1970s, techniques have been developed that allow scientists to cut, separate, and replicate DNA base-by-base. Using these tools, scientists can read the base sequences in DNA from any cell.

- ▶ **Restriction enzymes** cut DNA into smaller pieces, called restriction fragments, which are several hundred bases in length. Each restriction enzyme cuts DNA at a different sequence of bases.
- ▶ **Gel electrophoresis** separates different-sized DNA fragments by placing them at one end of a porous gel, then applying an electrical voltage. The electrical charge moves the DNA.
- ▶ Using dye-labeled nucleotides, scientists can stop replication at any point along a single DNA strand. The fragments can then be separated by size using gel electrophoresis and “read,” base-by-base.

The Human Genome Project was a 13-year international effort to sequence all 3 billion base pairs in human DNA and identify all human genes. The project was completed in 2003.

- ▶ The researchers identified markers in widely separated strands of DNA.
- ▶ They used “shotgun sequencing,” which uses a computer to match DNA base sequences.
- ▶ To identify genes, they found promoters, exons, and other sites on the DNA molecule.
- ▶ To locate and identify as many haplotypes (collections of linked single-base differences) in the human population as possible, the International HapMap Project began in 2002.
- ▶ The Human Genome Project identified genes associated with many diseases and disorders. From the project came the new science of **bioinformatics**, the creation and use of databases and other computing tools to manage data. Bioinformatics launched **genomics**, the study of whole genomes.
- ▶ The human genome project pinpointed genes and associated particular sequences in those genes with numerous diseases and disorders. It also found that the DNA of all humans matches base-for-base at most sites, but can vary at 3 million sites.
- ▶ The 1000 Genomes Project, launched in 2008, will catalogue the variation among 1000 people.

Chapter 14 The Human Genome Answers

Sandra J. Judd



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Genomes 4 T. A. Brown, 2018-12-07 Genomes 4 has been completely revised and updated It is a thoroughly modern textbook about genomes and how they are investigated As with Genomes 3 techniques come first then genome anatomies followed by genome function and finally genome evolution The genomes of all types of organism are covered viruses bacteria fungi plants and animals including humans and other hominids Genome sequencing and assembly methods have been thoroughly revised including a survey of four genome projects human Neanderthal giant panda and barley Coverage of genome annotation emphasizes genome wide RNA mapping with CRISPR Cas 9 and GWAS methods of determining gene function covered The knowledge gained from these techniques forms the basis of the three chapters that describe the three main types of genomes eukaryotic prokaryotic including eukaryotic organelles and viral including mobile genetic elements Coverage of genome expression and replication is truly genomic concentrating on the genome wide implications of DNA packaging epigenome modifications DNA binding proteins non coding RNAs regulatory genome sequences and protein protein interactions Also included are applications of transcriptome analysis metabolomics and systems biology The final chapter is on genome evolution focusing on the evolution of the epigenome using genomics to study human evolution and using population genomics to advance plant breeding Established methods of molecular biology are included if they are still relevant today and there is always an explanation as to why the method is still important Each chapter has a set of short answer questions in depth problems and annotated further reading There is also an extensive glossary Genomes 4 is the ideal text for upper level courses focused on genomes and genomics **Essential Genetics** , *Genomes 5* Terry A. Brown, 2023-04-28 Genomes 5 has been completely revised and updated It is a thoroughly modern textbook about genomes and how they are investigated As with previous Genomes editions techniques come first then genome anatomies followed by genome function and finally genome evolution The genomes of all types of organism are covered viruses bacteria fungi plants and animals including humans and other hominids Genome sequencing and assembly methods have been thoroughly revised to include new developments in long read DNA sequencing Coverage of genome annotation emphasizes genome wide RNA mapping with CRISPR Cas 9 and GWAS methods of determining gene function covered The knowledge gained from these techniques forms the basis of the chapters that describe the three main types of genomes eukaryotic prokaryotic including eukaryotic organelles and viral including mobile genetic elements Coverage of genome expression and replication is truly genomic concentrating on the genome wide implications of DNA packaging epigenome modifications DNA binding proteins non coding RNAs regulatory genome sequences and protein protein interactions Also included are examples of the applications of metabolomics and systems biology The final chapter is on genome evolution including the evolution of the epigenome using genomics to study human evolution and using population genomics to advance plant breeding Established methods of molecular biology are included if they are still relevant today and there is always an explanation as to why the

method is still important Genomes 5 is the ideal text for upper level courses focused on genomes and genomics Key Features A highly accessible and well structured book with chapters organized into four parts to aid navigation Superb artwork illustrates the key concepts and mechanisms Each chapter has a set of short answer questions and in depth problems to test the reader s understanding of the material Thoroughly up to date with references to the latest research from the 2020s

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