KAPITAŁ LUDZKI
NARODOWA STRATEGIA SPÓJNOŚCI

UNIA EUROPEJSKA
EUROPEJSKI
FUNDUSZ SPOŁECZNY

"PWP: Uniwersytet Jutra: Umiędzynarodowienie szkolenia w Uniwersytecie Gdańskim poprzez współpracę z Uniwersytetem Houston-Downtown"
Projekt współfinansowany ze środków Unii Europejskiej w ramach Europejskiego Funduszu Społecznego

Introduction to Human Genetics

Module 5 Human genomics

Web resources

UNIWERSYTET GDAŃSKI

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The Gene Gateway Workbook

A collection of activities introducing new users to the web resources that scientists access to learn about genetic disorders, genes, and proteins.

Using hereditary hemochromatosis as a model, access a variety of websites and databases to

- Learn about a genetic disorder and its associated gene.
- Identify mutations that cause the disorder.
- Find the gene on a chromosome map.
- Examine the gene's sequence and structure.
- Access the amino acid sequence of a gene's protein product.
- Explore the 3-D structure of the gene's protein product.

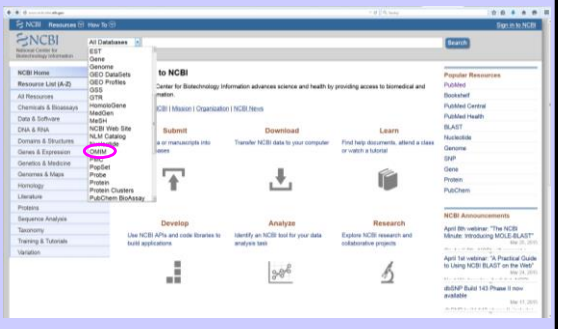
Why Use Hereditary Hemochromatosis as a Model?

- Hereditary hemochromatosis, a disorder in which too much iron accumulates in certain tissues and organs, is caused by changes in the DNA sequence of a single gene, so the genetic basis of this condition is easier to understand than more complex disorders caused by alterations in multiple genes.
- The gene and its protein product are relatively well studied. Three-dimensional structures of the protein product are available in PDB, the international repository for macromolecular structure data.
- Hereditary hemochromatosis is the most common autosomal recessive disorder affecting individuals of Northern European descent (about 1 in 200 Caucasians develop hereditary hemochromatosis).
- Effective methods for treatment are available with early diagnosis.

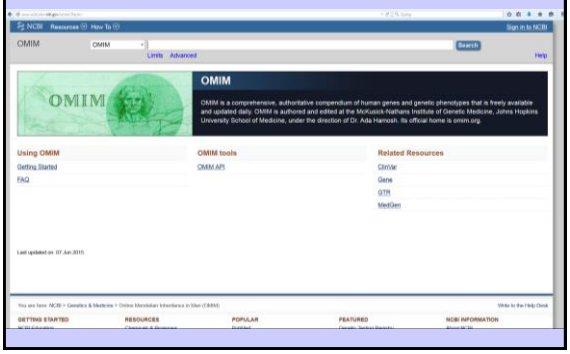
Some Basic Concepts to Understand Before Starting

- Genes are the basic physical and functional units of heredity. Each gene is located on a particular region of a chromosome and has a specific ordered sequence of nucleotides (the building blocks of DNA).
- Central dogma of molecular biology: DNA → RNA → Protein
  - Genetic information is stored in DNA.
  - Segments of DNA that encode proteins or other functional products are called genes.
  - Gene sequences are transcribed into messenger RNA intermediates (mRNA).
  - mRNA intermediates are translated into proteins that perform most life functions.
- Eukaryotic genes have introns and exons. Exons contain nucleotides that are translated into amino acids of proteins. Exons are separated from each other by intervening segments of DNA called introns. Introns do not code for protein, and they are removed when eukaryotic mRNA is processed. Exons are spliced back together to form the intron-free mRNA strand that is used as a template to make proteins.
- Special cellular components (ribosomes) use the triplet genetic code to translate the nucleotides of an mRNA sequence into the amino acid sequence of a protein. A Table of Standard Genetic Code is provided on page 50 of this workbook.
- There are 20 different amino acids. Proteins are created by linking amino acids together in a linear fashion to form polypeptide chains. See the Table of Standard Genetic Code on page 50 for single-letter and three-letter abbreviations for the 20 different amino acids.
- Polypeptide chains fold into 3-D structures that can associate with other molecular structures to perform specific functions.

Activity 1

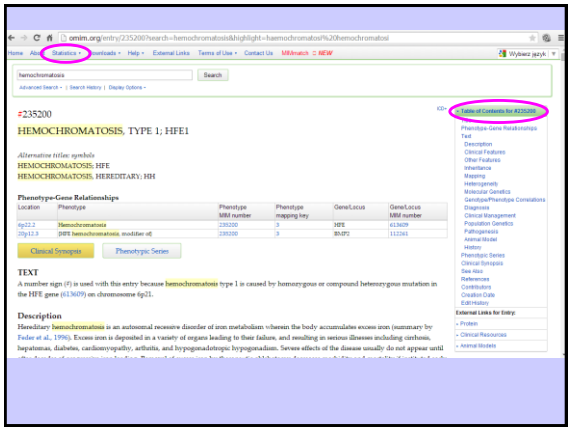


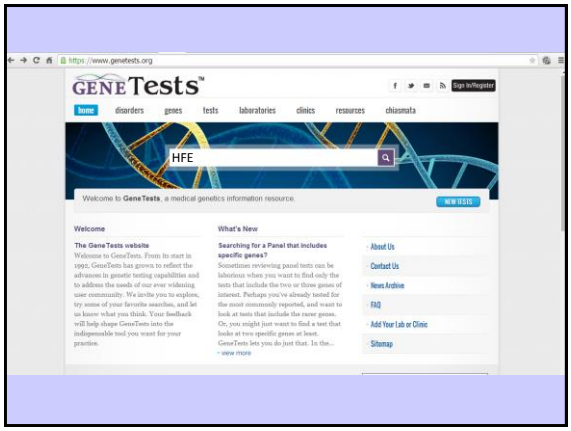
Activity 1

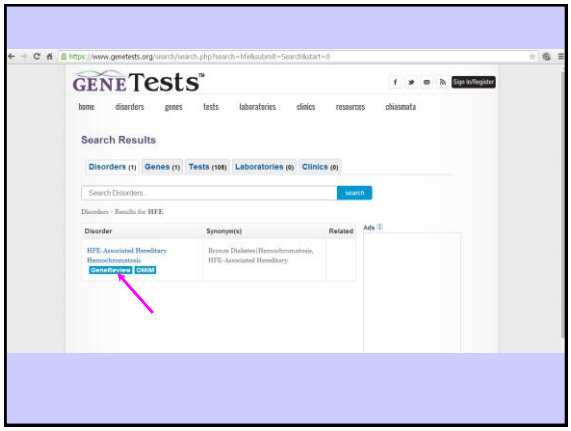


Activity 1

www.omim.org







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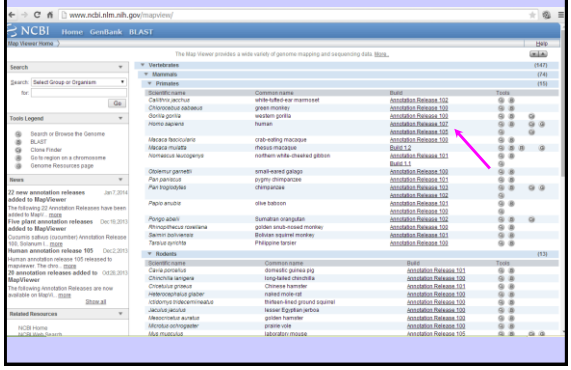

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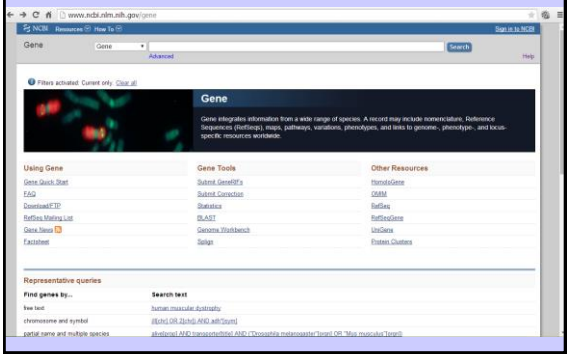
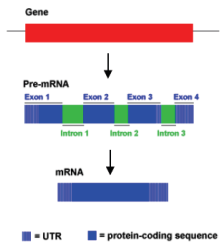


What's the difference between exons and coding sequence?

Exons often are described as short segments of protein coding sequence. This is a bit of an oversimplification. Exons are segments of sequence spliced together after introns have been removed from pre-mRNA. Exons carry the coding sequence of a gene, but some exons may contain no coding sequence. Portions of exons or even entire exons may contain sequence that is not translated into amino acids. These are the untranslated regions (UTR) of mRNA. UTRs are found upstream and downstream of the protein-coding sequence. See diagram on right.



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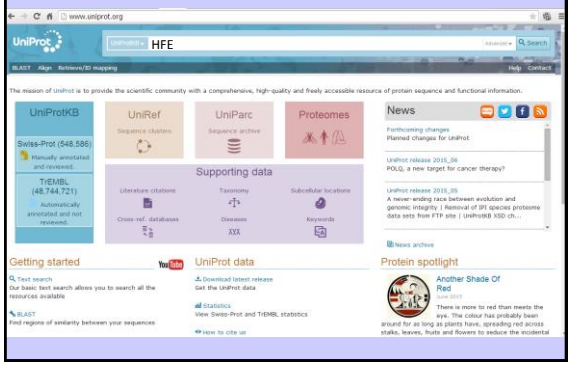

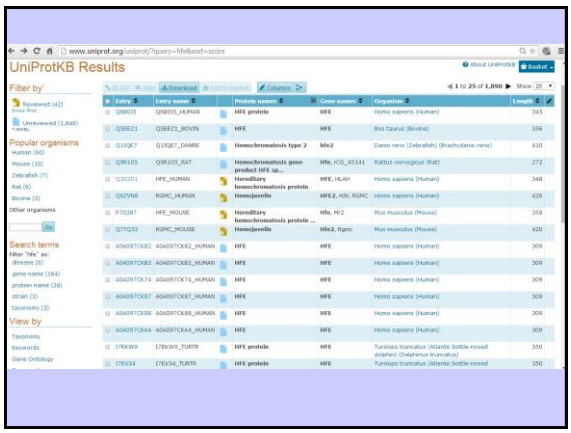

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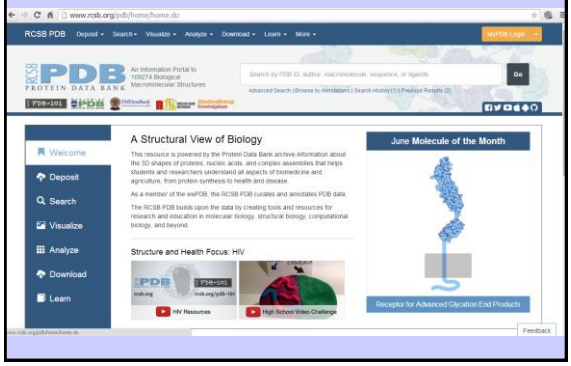
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