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DZIAŁALNOŚĆ FUNDUSZU SPOŁECZNEGO

"WFB: Uniwersytet Jafra - Uprzodkowienie kultury i wiedzy. Główne poparcie wspólnocie z Uniwersytetem Houston-Oriented".
Projekt współfinansowany ze środków Unii Europejskiej w ramach Europejskiego Funduszu Spójności

Introduction to Human Genetics

Module 5 Human genomics

Web resources

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.

- 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



The screenshot shows the OMIM (Online Mendelian Inheritance in Man) homepage. At the top, there is a search bar with the placeholder "Search OMIM". Below the search bar, there are several links: "About OMIM", "Resources", "How To Use OMIM", "OMIM", "OMIM-MI", "Limits", "Advanced", "Search", and "Help". The main content area features a large green circular graphic with the text "OMIM" and a portrait of a classical figure. To the right of the graphic, the text "OMIM is a comprehensive, authoritative compendium of human genes and genetic phenotypes that is freely available and updated daily. OMIM is authored and edited at the McKusick-Nathans Institute of Genetics, Johns Hopkins University School of Medicine, under the direction of Dr. Ada Hamosh. Its official home is [omim.org](#).

Using OMIM

- Getting Started
- FAQ

OMIM tools

- OMIM API

Related Resources

- OMIM
- Gene
- GTEx
- MedGen

Last updated on: 07 Jun 2015

You are here: NCBI > Genetics & Medicine > Online Mendelian Inheritance in Man (OMIM)

Help | To the Help Desk

SETTINGS STARTED RESOURCES POPULAR FEATURED NCBI INFORMATION

Activity 1

www.omim.org

OMIM® Online Mendelian Inheritance in Man®
An Online Catalog of Human Genes and Genetic Disorders
Updated 2 June 2015

Search OMIM... Advanced Search | OMIM, Clinical Syndromes, Gene Map | Search History
Need help? | Example Searches, OMIM Search Help, OMIM Tutorial
Mirror sites | us-east.omim.org, europe.omim.org

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OMIM® Online Mendelian Inheritance in Man®
An Online Catalog of Human Genes and Genetic Disorders
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Search OMIM... Advanced Search | OMIM, Clinical Syndromes, Gene Map | Search History
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235200 HEMOCHROMATOSIS, TYPE 1; HFE1
Alternative title: symbols
HEMOCHROMATOSIS, HFE
HEMOCHROMATOSIS, HEREDITARY; HH

Phenotype-Genotype Relationships

Location	Phenotype	Phenotype MIM number	Phenotype Mapping key	Gene/Locus	Gene/Chromosome MIM number
6p21.2	Hemochromatosis	235200	3	HFE	613609
2p13.3	DHF hemochromatosis, modifer q	235200	3	HMP2	111281

Clinical Synopsis **Phenotypic Series**

TEXT
A question mark (?) is used with this entry because hemochromatosis type 1 is caused by hemozygous or compound hemozygous mutations in the HFE gene (613609) on chromosome 6p21.

Description
Hereditary hemochromatosis is an autosomal recessive disorder of iron metabolism wherein the body accumulates excess iron (summary by Feder et al., 1996). Excess iron is deposited in a variety of organs leading to their failure, and resulting in serious illnesses including cirrhosis, hepatoma, diabetes, cardiomyopathy, arthritis, and hypogonadotropic hypogonadism. Severe effects of the disease usually do not appear until

Table of Contents for 235200

- Phenotype-Genotype Relationships
- Text
- Description
- Clinical Features
- Phenotypic Variants
- inheritance
- Hemochromatosis
- Molecular Genetics
- Gene/Chromosome
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- Diagnosis
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- Clinical Synopsis
- Phenotypic Series
- References
- Creation Date
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- Protein
- Protein Resources
- Animal Models

GENE Tests™

Welcome to GeneTests, a medical genetics information resource.

What's New
The GeneTests website has undergone a facelift. From its start in 1997, GeneTests has grown to reflect the advances in genetic testing capabilities and to address the needs of the medical genetics user community. We invite you to explore, sometimes revisiting panellets to be laboratories when you want to find only the most up-to-date panellets for a specific test of interest. Perhaps you've already tested for the most commonly reported, and least likely to be clinically relevant variants. Or, you might just want to find a test that looks at two specific genes at least. GeneTests let you do just that. In the... view more

Searching for a Panel that includes specific genes?
Sometimes reviewing panellets can be laborious when you want to find only the most up-to-date panellets for a specific test of interest. Perhaps you've already tested for the most commonly reported, and least likely to be clinically relevant variants.

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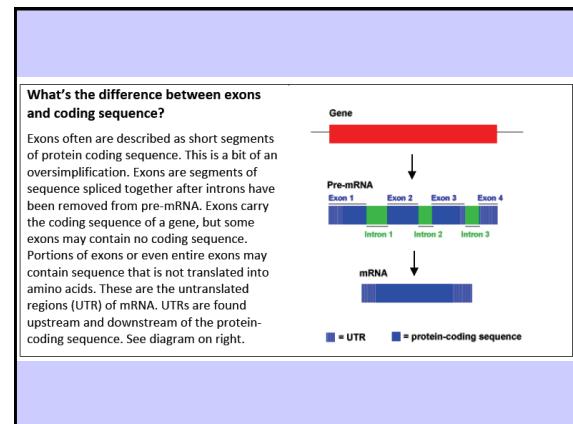


Search Results

Disorder	Synonym(s)	Related
HFE-Associated Hereditary Hemochromatosis	Bronze Disease; Hemochromatosis, HFE-Associated Hereditary	Ads

Activity 3

The screenshot shows the NCBI Gene homepage with the URL www.ncbi.nlm.nih.gov/gene. The search bar at the top contains the query 'C. elegans'. Below the search bar, the results are displayed under the heading 'Gene' with a 'Search' button. The results include a summary box for 'C. elegans' with a 'View details' link, a 'Gene structure' section with a diagram, and a 'Gene expression' section with a heatmap. A sidebar on the left lists 'Using Gene' resources like GeneQuick, EAF, DiseaseID, RefSeq, and GeneTests, along with 'Facets' for search filtering. A 'Gene Tools' sidebar on the right includes links for Submit GeneID's, Submit Corrective, Status, BLAST, and Genome ToolsSearch. A 'Other Resources' sidebar on the right includes links for HomoloGene, OMIM, Reference, UniGene, and Patent Clusters. At the bottom, a 'Representative queries' section lists 'Find genes by...' (taxon, chromosome, and symbol, protein name and multiple species), a 'Search text' input field with 'Human muscular dystrophy' entered, and a 'Search' button.



Activity 4



The mission of UniProt is to provide the scientific community with a comprehensive, high-quality and freely accessible resource of protein sequence and functional information.

UniProtKB

Swiss-Prot (548,596)
Manually annotated and reviewed.

TrEMBL (441,721)
Automatically annotated and not reviewed.

UniRef
Sequence clusters

UniParc
Sequence archive

Proteomes
Protein families

News
Funding changes
Planned change for UniProt

Supporting data

Liberation rotations

Cross ref. databases

Proteome

Proteome

Subcellular locations

Keywords

Getting started

UniProt search
Our basic text search allows you to search all the resources available

BLAST
Find regions of similarity between your sequences

UniProt data

Download latest release
Get the UniProt data

Statistics
View Swiss-Prot and TrEMBL statistics

How to cite us

Protein spotlight
Another Shade Of Red

There is more to red than meets the eye. The colour has probably been around for as long as plants have, spreading red across stable, leaves, fruits and flowers to reduce the ecological

Activity 5



The screenshot shows the homepage of the RCSB PDB (Protein Data Bank) website. The top navigation bar includes links for Deposit, Search, Visualize, Analyze, Download, Learn, and More. A search bar is present, along with a "MyPDB Login" button. The main content area features a "June Molecule of the Month" section with a 3D molecular model and a "Structure and Health Focus: HIV" section with links to "IPMB-101", "IPMB-102", "IPMB-103", "IPMB-104", "IPMB-105", "IPMB-106", and "IPMB-107". A sidebar on the left provides links for Welcome, Deposit, Search, Visualize, Analyze, Download, and Learn.



<https://www.flickr.com/photos/fleur-design/2957704923/sizes/o/>