WEB RESOURCES

1. Hello! Welcome to the fifth Module of Introduction to Human Genetics. This is the second lecture in this module and here I'm going to encourage you to use online data sources from the Human Genome Project to investigate any human gene or genetic disorder of your interest.

2. In the supplementary materials for this module you can find a pdf file with The Gene Gateway Workbook. It's an excellent guide to get started using bioinformatics databases. It's a set of five activities with screenshots and step-by-step instructions designed to introduce new users to genetic-disorder and bioinformatics resources freely available on the Web. Since it was last updated February 2011, you'll see that in the meantime some of the presented websites have changed their interfaces; nevertheless, following the instructions from the workbook you should still be able to navigate through them.

3. You'll be asked to perform all five activities for one model genetic disorder - hereditary hemochromatosis. In the introduction to the workbook you'll find the reasons for choosing this disease as a model...

4. ... as well as basic knowledge you should possess before starting. Please, now pause the video to study it carefully. Ok, now you're ready to start.

5. In the first activity you'll learn about the genetic disorder, its associated gene and mutations. The first database you'll go to is the Online Mendelian Inheritance in Man (OMIM).

6. You may access it via the NCBI home page as it is shown in the workbook or

7. directly by typing www.omim.org in the address bar.

8. If you find the instructions from the workbook insufficient you might also wish to go through the OMIM tutorials which are available here.

9. Type ...

10. ...the name of the disease you are interested in in the search box.

11. The site with the result will look a bit different than a screenshot presented in the workbook on page 8, but if you compare their tables of contents – these are pretty much the same and you should be able to follow the instructions from the workbook.
When it comes to the question about the number of genes described in the OMIM – you will find the answer selecting Statistics tab.

12. Next website shown in the workbook is GeneTests. Again its home page has changed since February 2011 but if you type the gene symbol HFE in the navigation box that will lead you to...
13. ... the search result page from which you can go to GeneReview that is presented in the workbook.

14. In the second Activity you will navigate through the NCBI Map Viewer. It provides access to several different types of maps for different organisms. You will work on a human chromosome map. If you haven't done Module 2 of our course yet, perhaps after this activity you'd like to go there and learn more about chromosomes and cytogenetics.
The NCBI Map Viewer website itself hasn't changed much but the builds have been updated so you will see different annotation release from the one in the workbook. Nonetheless, you should be able to do everything you are asked for, following instructions from the workbook.

15. Similarly, Gene and GeneBank resources from the third Activity haven't changed much. Only the data has been updated. And you should have no problems to perform this activity. There is one thing in this activity I want to draw your attention to.

16. The explanation of the difference between exons and coding sequence. Please, acquaint yourself with this distinction.

17. In the Activity 4 you'll go to the UniProt Protein Knowledgebase. You'll notice that UniProt website has changed its appearance but once you -> start your search...

18. ... the results will look very similar to what we have in the workbook. So just follow the instructions from the workbook to complete this activity.

19. In the last fifth Activity you'll explore the sequence and structure of the gene's protein product. Go to the Protein Data Bank and follow the instructions from the workbook.

20. I hope you'll have much fun surfing human genome on the Internet.