

Exercises for OMIM

(Version 7, 2012)

You can choose to do these using the questions as your only guide—or see the following pages for the step-by-step checklist to finding these answers.

1) You are interested in determining if there are any phenotypes associated with the human RANKL gene, and whether this association is due to variation in the gene. Do a basic search for the gene RANKL, also known as TNFSF11. What is the genetic location of the gene? What disease is associated with the gene? Can you quickly locate other reports that offer information on phenotypes that might be similar to yours?

Skills: Basic OMIM Search; Understanding Your Results; Gene Entries, Phenotype Entries, Phenotypic Series.

2) Imagine that you have a patient displaying evidence of myopathy that has been linked to the chromosomal location 2p13. Conduct a Gene Map search for the area and assess phenotypes in the region that may be the cause..

Skills: Gene Map Searches, Customizing Displays, Linking to Gene and Phenotype Entry Reports from Results lists.

3) Explore the utility of different formatting and search options for focusing your searches using various searches for rheumatoid arthritis.

Skills: Search Formatting, Advanced Searches.

**OMIM Exercises, version 7,
Corresponds to the data available in November 2012**

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Step-by-Step checklist/instructions for the OMIM introduction exercises

1) You are interested in determining if there are any phenotypes associated with the human RANKL gene, and whether this association is due to variation in the gene. Do a basic search for the gene RANKL, also known as TNFSF11. What is the genetic location of the gene? What disease is associated with the gene? Can you quickly locate other reports that offer information on phenotypes that might be similar to yours?

Step	Action	✓
1	Go to the OMIM homepage at http://www.omim.org .	
2	In the basic search box, enter the text: RANKL , and then click "Search".	
3	Examine your results. The top result is a gene, as denoted by the asterisk, but uses TNFSF11 as the gene name rather than RANKL . <i>Note authors may use synonyms rather than primary names but OMIM searches still find the correct entry.</i>	
4	Click the 602642 TUMOR NECROSIS FACTOR...TNFSF11 link.	
5	Scroll through the gene entry page and notice the detailed information available about the gene. <i>Note that the genetic location of this gene is 13q14.11.</i>	
6	Locate the Table of Contents link towards the upper right of the page, and click the link to open the Table of Contents . <i>Note that you can navigate to any section of the report using the links in the Table of Contents (TOC).</i>	
7	In the TOC, locate and click the Table View link found under the Allelic Variants link. <i>Note that the window reloads to display a table of allelic variants associated with osteopetrosis.</i>	
8	Click your browser back button to return to the gene page.	
9	At the top of the gene page, in the "Gene Phenotype Relationships" table, click the 259710 link under the "Phenotype MIM number" heading. <i>Note that the phenotypic series offers information on phenotypes that might be similar to yours, but that occur in other genomic locations.</i>	
10	Click open the phenotypic series and explore it, if you would like. <i>Note, you can see genomic locations for various osteopetrosis phenotypes, as well as other details.</i>	

Notes:

2) Imagine that you have a patient displaying evidence of myopathy that has been linked to the chromosomal location 2p13. Conduct a Gene Map search for the area and assess phenotypes in the region that may be the cause.

Step	Action	✓
1	Towards the top of any OMIM page, in the Advanced Search area, locate and click the “OMIM Gene Map” link. <i>Note that the Gene Map search form will open.</i>	
2	In the search box, enter the genomic location 2p13 , and then click “Search”. <i>Note that 10 results in the 2p13 area are displayed. We could have done a Gene Map search for the term “myopathy”, but our results would have been across many genomic locations and the display would begin with chromosome 1 results.</i>	
3	Although we could use the navigation buttons (Back3, Forward 3, etc.) to explore additional results, we will instead click the “Show 100” link.	
4	Scroll or use your browser’s Find function to locate the first gene or phenotype description that includes the term “myopathy” <i>Note the 604454 entry (WELANDER DISTAL MYOPATHY; WDM)</i>	
5	Examine the 604454 entry row for WELANDER DISTAL MYOPATHY. <i>Note both the gene and phenotype MIM numbers are the same.</i>	
6	Mouse over the 2 in the Pheno map key column to determine how this phenotype was placed on the map. <i>Note that WDM was mapped by statistical methods.</i>	
7	Scroll or use your browser’s Find function to locate the next gene or phenotype description that includes the term “myopathy” <i>Note that the Dysferlin gene is associated with a myopathy phenotype, as well as 2 muscular dystrophy phenotypes.</i>	
8	To learn more about the Dysferlin gene, under the Gene/Locus MIM number column, click the 603009 link. <i>Note that the *603009 Dysferin gene report opens.</i>	
9	Towards the top right of the report, click the Table of Contents (TOC) open. Use it to explore any type of gene information that interests you.	
10	To explore the phenotypes associated with the Dysferin gene, click any of the Phenotype MIM numbers located in the Gene Phenotype Relationships table.	

NOTES:

3) Explore the utility of different formatting and search options for focusing your searches using various searches for rheumatoid arthritis.

Step	Action	✓
1	Using any OMIM basic search box, enter the text: Rheumatoid Arthritis . Then click “Search” . <i>Note the number of results returned. This search is essentially for: Rheumatoid OR Arthritis.</i>	
2	Modify your search using any OMIM basic search box, by entering the text: -Rheumatoid Arthritis . Then click “Search” . <i>Note that fewer results are returned. This search can be translated as: Arthritis NOT Rheumatoid.</i>	
3	Leaving your search text in the search box, click any OMIM advanced search link .	
4	Ensure that your search text -Rheumatoid Arthritis remains in the search box. Then in the “Search In:” area, click the Title check box on . <i>Note that this will confine the search to text occurring in entry titles.</i>	
5	Click any Search button . <i>Note that there are many fewer results.</i>	
6	Initially the 186580 BLAU SYNDROME result may not appear to conform to our search criteria. To investigate, click on its link .	
7	On the 186580 entry page, examine the alternative titles area . <i>Note that arthritis is found in one of the alternative titles.</i>	
8	To explore additional search syntax available for focusing searches, mouse over the top “Help” tab , and click the “Search Help” link .	
9	Navigate to and read about any technique that interests you. <i>Note that there are many search formatting options that allow you to create elegant and focused searches.</i>	
10	Practice the syntax you have learned to perform searches that interest you . <i>Note that the Sample Search link on the OMIM homepage offers a few examples, with explanations of the search.</i>	

Notes: