

Challenge questions

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GTR

www.ncbi.nlm.nih.gov/gtr

1. Find information about the BRCA1 gene
 - a. What conditions are associated with the BRCA1 gene?
2. Find all tests for the BRCA1 gene in GTR
 - a. Scan the list and identify tests for the BRCA1 gene only vs. panels with BRCA1
 - b. Restrict your search to tests offered by US labs.
 - i. How many tests for BRCA1 are offered in the US?
 - ii. How many labs offer those tests?
 - iii. What are some differences between these tests?
3. Find practice guidelines for Hereditary breast and ovarian cancer syndrome (HBOC)
4. From GTR, find information about BRCA1 in ClinVar
5. Find information about cystic fibrosis in GTR
 - a. Find consumer-oriented information about cystic fibrosis to print out for your patient
 - b. From GTR, find information in ClinVar about variations in the CFTR gene

MedGen

<http://www.ncbi.nlm.nih.gov/medgen/>

6. Find a list of Professional Guidelines for genetics on the MedGen home page
 - a. Do you need to find these guidelines directly from MedGen's home page?
7. Find information about Duchenne muscular dystrophy
 - a. What is the mode of inheritance?
 - b. What is the gene? Which chromosome is that gene located on?
 - c. Open the GeneReview section on differential diagnosis.
 - d. How many genetic tests does GTR have for Duchenne muscular dystrophy?
 - e. Name three clinical features of Duchenne muscular dystrophy that affect the cardiovascular system

ClinVar

<http://www.ncbi.nlm.nih.gov/clinvar/>

8. Find DMD gene variants in ClinVar
9. What are the 2 most common types of DMD gene variation reported in ClinVar?

GeneReviews

<http://www.ncbi.nlm.nih.gov/books/NBK1116/>

10. Search for a *GeneReview* from the GTR home page
 - a. By condition name: Huntington disease
 - b. By gene name: COL1A1
 - i. COL1A1 does not cause all of these disorders. Why does 'Vascular Ehlers-Danlos syndrome' show in the result list (don't read the entire record to find out)?
 - c. By author: Braverman
 - i. How do you limit records to the author's name vs. the name is somewhere in the record (e.g. as a citation)?

Medical Genetics Summaries

Genetic variants and drug responses

<http://www.ncbi.nlm.nih.gov/books/NBK109194/>

11. Find the drug response record in MedGen for Prilosec
 - a. What gene is an important determinant of variation in drug metabolism?
 - b. Find all genetic tests in GTR for omeprazole response
 - i. What information might you use in choosing a test?
 - c. Find the Therapeutic recommendations section (go back to MedGen)
 - d. Find the specific Medical Genetics Summary (go back to MedGen)