

Assignment 3. Due Wednesday 19 Jan There is a group of genetic disorders called trinucleotide repeat disorders (https://en.wikipedia.org/wiki/Trinucleotide_repeat_disorder). A large subset of these involve repeats of CAG that encode stretches of glutamine, which has amino acid symbol Q. So these are sometimes called poly-Q diseases.

Please hand in a hard-copy document with two screenshots as follows. You can consult with / get help from other class members, but you cannot copy their screenshots. Please send in your own screenshots.

Find the gene ATXN3 in the UCSC genome browser.

Please be sure to use the hg19 reference genome. To do this go to http://genomeasia.ucsc.edu/cgi-bin/hgGateway?hgsid=741515590_lwevGa81AEJZBs3CgBH3PRmslFKE and use the “Genomes” pull down to select this genome

Type ATXN3 in the search bar labeled “enter position, gene symbol ...”

This will take you to a list of annotated transcripts for the gene.

Click on the first one:

`“ATXN3 (uc001yac.4) at chr14:92524896-92572965 - Homo sapiens ataxin 3 (ATXN3), transcript variant reference, mRNA.”`

which will take you back to the graphical display.

Under the graphical display there will be a bewildering variety of selection boxes. Set the following

“Mapping and Sequencing” -> Base Position (full)
“Genes and Gene Predictions” -> NCBI RefSeq (dense)
“Phenotype and Literature” -> ClinVar Variants (pack)

tracks **and turn off (“hide”) all other tracks (then click “refresh”)**

Question 1: *** Make a screen shot and paste it in a document to turn in as hard copy

Find the poly Q / CAG repeat, which should be obvious in the ClinVar Short Variants track and in the “Clinvar interp” track (red dot on the “P” = pathogenic) line.

Zoom it to the poly-Q region by selecting the nearby region at the “ruler” at the top of the graphical display.

Click the “reverse” button just underneath the graphical display.

Then you can see the Poly Q in one of the translation frames

Question 2 Take a screenshot of the graphical display region and put it in the document. Make sure it is big enough to read the DNA sequence.

Write down the genomic coordinates of the current view roughly centered on the poly-Q repeat

Question 3 Turn off the “reverse” button. The CAGCAGCAG... Repeats encoding the poly Q are now what (at the DNA level)?