

IGV assignment due Monday 24 Jan

Open the IGV web app: <https://igv.org/app/>

Under “Genome” select hg19 (upper far left pull down)

Under tracks select “Platinum”, then NA12878

(FYI, info on the “Platinum” genomes is at <https://www.illumina.com/platinumgenomes.html>)

Under tracks, select “Annotations”, then “Repeat Masker” (to show the Repeat Masker track)

Go to: chr14:92,537,279-92,537,450

(This is the poly-Q region in ATXN3)

At the gear icon to the right of the nucleotide sequence, select “Three-frame translation” and “Reverse”; this “Reverse” refers to the translation, not the sequence.

Click on the Repeat Masker rectangle. What kind of repeat is this?

Take a screenshot and include it a printout.

For your reference, here is a different part of the ATXN3 gene, which a different Repeat Masker region shown (this repeat is an Alu, a specific subtype called AluSx1). The

