Clinical and Research Genomics Assignment #5

Complex Genome Re-arrangements, Transposons, and Genetic Variant Calling

---

Assignment: Answer questions on genetic variation and clinical genomics.
Due before 5/13/22 5:00PM

---

1. How is the positive strand of a chromosome defined?

2. What are the differences between indels, structural variants (SVs), and copy number variants (CNVs)? Why is this a useful distinction to make for both detection purposes and general biology?

3. Where in terms of chromosome and strand would each of a pair of DNA WGS short reads align if they flanked (but did not span) the breakpoint of a fusion joining gene A on the negative strand of chr 1 to gene B on the positive strand of chr 2?

4. In his presentation, Dr. Snyder presented a case where his team was able to use a smartwatch to identify a leading indicator of COVID-19 infection. This is your opportunity to consider how you might design a study that would use a smart watch or other personal device to collect data that could be used to correlate to a molecular omics (expression, metabolome, proteome, etc) biomarker. For example: fertility cycles, vertigo, or weight loss/gain. There is no right or wrong answer, but we are keen to see how you choose to develop your selected experimental design including hardware, software, and selected biomarkers and the ways you would choose to track them.

   Link to Dr. Snyder COVID-19 smartwatch app paper:
   https://www.nature.com/articles/s41551-020-00640-6

Please hand the assignment on the day of the lecture, or email if you cannot attend.
For any questions, please contact Chandrima Bhattacharya (chb4004@med.cornell.edu), Evan Afshin (eba2001@med.cornell.edu), or Professor Mason (chm2042@med.cornell.edu)