ANGSD 17 March, 2021

Calling single nucleotide variants and structural variants

Single Nucleotide Variants (SNVs)

1. What are the differences between whole exome sequencing and whole genome sequencing for SNVs?
2. Assuming that a region is sequenced using both methods, do they always detect the same mutations?
3. Import/open the attached vcf file:
    a. What is the TCGA study ID for this sample?
    b. What genome build was this sample aligned to?
4. Are all of the detected mutations "real"?

Structural variants (SVs)

1. What is the major difference on the impact on the genome between SVs and SNVs?
2. What sequencing approach is suitable for SVs calling?
3. Can we detect all possible SVs?

Email your responses to Andre Forbes (anf2034@med.cornell.edu) and Alex Fundichely (alm2069@med.cornell.edu) by midnight on Tuesday 16 March 2021.