

# Prioritization of structural variants based on known biological information

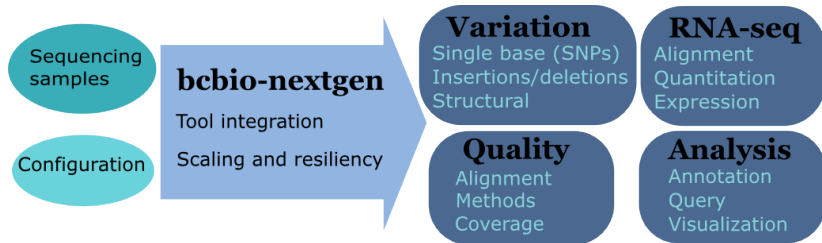
Brad Chapman

Bioinformatics Core, Harvard Chan School

<http://j.mp/bcbiolinks>

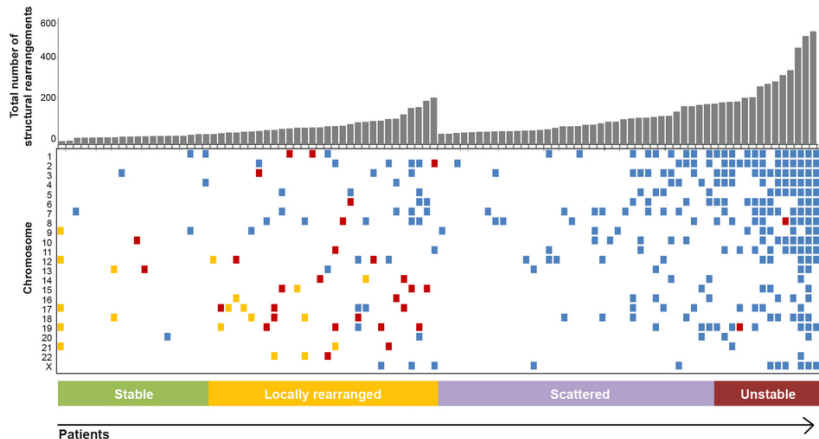
10 July 2015

# Community developed variant calling



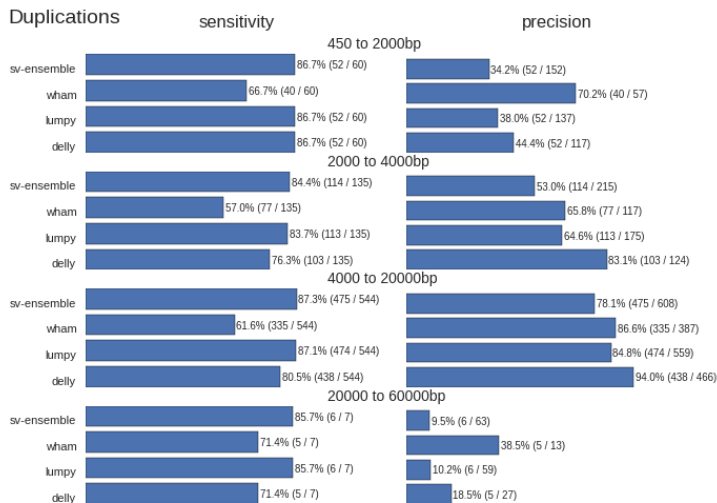
<https://github.com/chapmanb/bcbio-nextgen>

# Structural changes critical in cancer



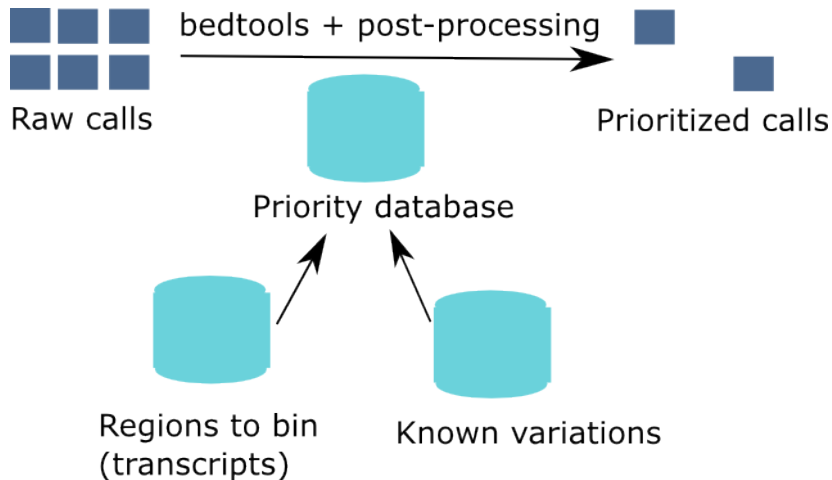
<http://www.nature.com/nature/journal/v518/n7540/full/nature14169.html>

# Structural variants hard to detect



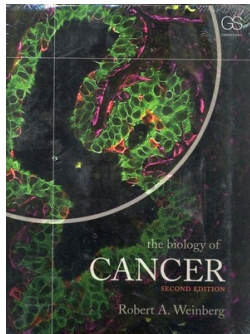
<http://bcb.io/2015/03/05/cancerval/>

# Prioritize variants in known regions



# Public cancer variant databases

- CIViC: <https://civic.genome.wustl.edu>
- IntOGen: <http://www.intogen.org>



<http://www.amazon.com/The-Biology-Cancer-Robert-Weinberg/dp/0815340761>

# Prioritization – summary

- Structural variants critical but hard
- Use known biological information
- Support community databases
- bcbio.prioritize  
<https://github.com/chapmanb/bcbio.prioritize>
- bcbio  
<https://github.com/chapmanb/bcbio-nextgen>