INDELs as genetic markers for local adaptation

Samuel Perini et al.

6th September 2019

# Background

1. Mutations are changes in the nucleotide sequence of the DNA and without them evolution will not exist because there will be no genetic variation for genetic drift and natural selection to act upon. Mutations occur in different sizes and as they go from one base to (size of the longest variant), their contribution to genetic diversity increases as well as their potential to decrease fitness of the individuals carrying the mutation. Accordingly, different species and also alternative forms of the same species (ecotypes) have been found to differ for a greater number of single nucleotides than for long structural variants. The explanation behind this observation is almost intuitive, large modifications to a functional system are more likely to cause damage than small ones (**???**). However, there are examples of structural variants with positive effects such as …  
   Originally, mutations were detected using (first method of variant calling) and now, after the emergence of high-throughput sequencing technologies, it is affordable to scan whole genomes in search for different types of mutations. Single nucleotide polymorphisms (SNPs) have been primary targets of genetic and genomic analyses that aimed to (applications for SNPs (Chen et al. 2009)). Few studies have expanded their analysis to other types of genetic variants in order to find additional patterns of evolutionary change. For example, short insertions and deletions (INDELs 50 bp) (Chen et al. 2009; Barton and Zeng 2019)… The notion about the importance of INDELs is not new in the field of evolutionary genetics but our knowledge about their role is still limited to a few model species.
2. Low number of studies has used INDELs because of the challenges associated with INDEL discovery (Narzisi and Schatz 2015).
3. INDELs can show a different and complementary evolutionary pattern compared to the one based on SNPs.
4. SNP-based study in *Littorina saxatilis*, species biology and results.

# Questions

Can short INDELs be used as genetic markers for patterns of local adaptation? How does INDEL variation differ from SNP variation in *L. saxatilis*?

# Hypothesis

Nucleotide divergence between closely related species can be composed of a higher percentage of short INDELs than SNPs (Britten 2002) and the same pattern might be found between two locally adapted populations of the same species. Detecting INDELs can reveal additional evolutionary processes that would otherwise be unnoticed by a SNP-based analysis.

# References

Barton, Henry J, and Kai Zeng. 2019. “The Impact of Natural Selection on Short Insertion and Deletion Variation in the Great Tit Genome.” *Genome Biology and Evolution* 11 (6): 1514–24.

Britten, Roy J. 2002. “Divergence Between Samples of Chimpanzee and Human Dna Sequences Is 5%, Counting Indels.” *Proceedings of the National Academy of Sciences* 99 (21): 13633–5.

Chen, Jian-Qun, Ying Wu, Haiwang Yang, Joy Bergelson, Martin Kreitman, and Dacheng Tian. 2009. “Variation in the Ratio of Nucleotide Substitution and Indel Rates Across Genomes in Mammals and Bacteria.” *Molecular Biology and Evolution* 26 (7): 1523–31.

Narzisi, Giuseppe, and Michael C Schatz. 2015. “The Challenge of Small-Scale Repeats for Indel Discovery.” *Frontiers in Bioengineering and Biotechnology* 3: 8.