

Lung Pre-cancer

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1 Introduction

1.1 Lung Cancer

1.2 Precancer

1.3 Study Objectives

2 Materials

2.1 List of IPNs

2.1.1 Carcinoma *in situ*

Carcinoma *in situ* (CIS)

2.1.2 Adenocarcinoma *in situ*

Adenocarcinoma *in situ* (AIS)

2.1.3 Atypical Adenomatous Hyperplasia

Atypical adenomatous hyperplasia (AAH)

2.1.4 Dysplasia

2.1.5 Minimally Invasive Adenocarcinoma

Minimally invasive adenocarcinoma (MIA)

2.2 Data Structure & Count

3 Methods

3.1 Workflows

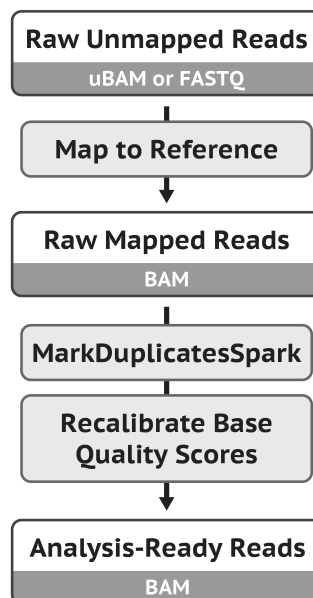


Figure 1: Workflow for data pre-processing for variant discovery (Van der Auwera et al., 2013; DePristo et al., 2011)

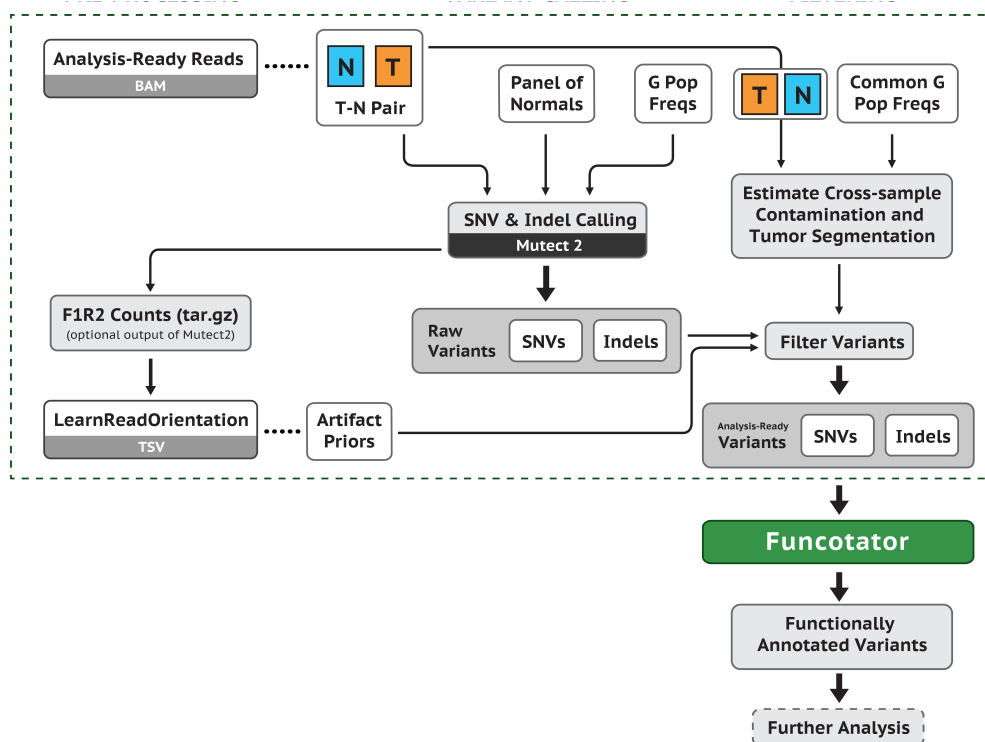


Figure 2: Somatic short variant discovery workflow (Van der Auwera et al., 2013; DePristo et al., 2011)

4 Results

4.1 Quality Check with FastQC

4.2 Quality Check with Sequenza

4.3 Progressive Evolution at Single Nucleotide Level

4.4 Macro-evolution at Chromosomal Level

4.5 Selective Clonal Sweep during Neoplastic Evolution

4.6 Cancer Gene Mutation during Cancer Evolution

4.7 Distinct Drivers and Genetic Constraints in Multifocal IPNs

5 Discussion

6 References

- DePristo, M. A., Banks, E., Poplin, R., Garimella, K. V., Maguire, J. R., Hartl, C., ... others (2011). A framework for variation discovery and genotyping using next-generation dna sequencing data. *Nature genetics*, 43(5), 491.
- Van der Auwera, G. A., Carneiro, M. O., Hartl, C., Poplin, R., Del Angel, G., Levy-Moonshine, A., ... others (2013). From fastq data to high-confidence variant calls: the genome analysis toolkit best practices pipeline. *Current protocols in bioinformatics*, 43(1), 11–10.

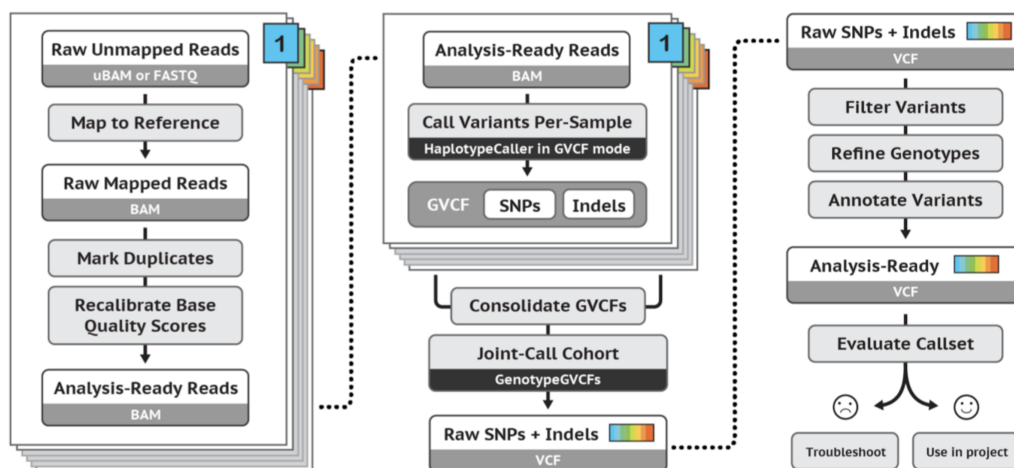


Figure 3: Germline short variant discovery workflow (Van der Auwera et al., 2013; DePristo et al., 2011)

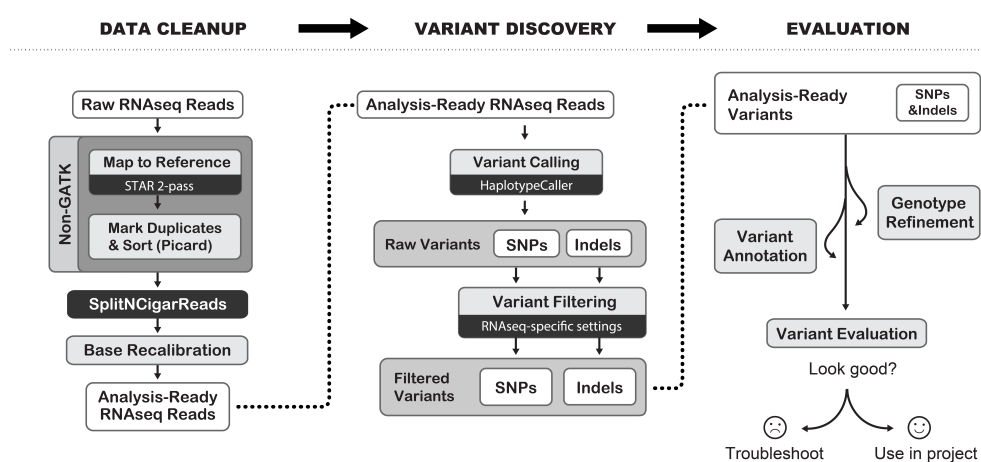


Figure 4: RNA-seq short variant discovery workflow (Van der Auwera et al., 2013; DePristo et al., 2011)

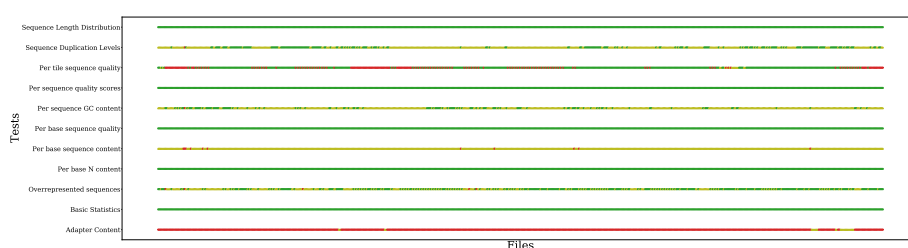


Figure 5: FastQC results with WES data

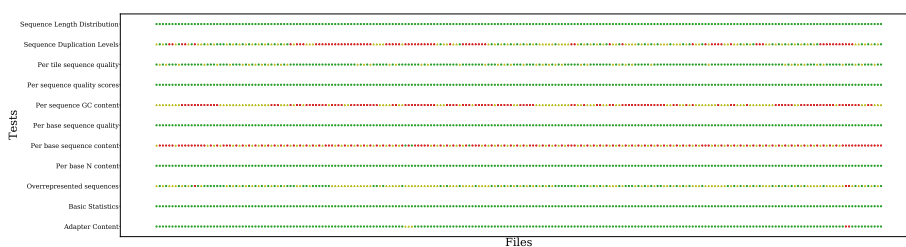


Figure 6: FastQC results with WTS data