Lung Cancer

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Overview

Introduction

Objectives

- Find different mutations
  - between WES
  - between WTS
  - from cancer
  - from pre-cancer
- Pathway examine from the mutations
  - of WES
  - of RNA-seq
- Ultra-deep sequencing to find an *infinitesimal* quantity of Non-Circulating Tumor DNA
  - from blood
  - from urine
  - frrom bronchus
- Diagnostic performace

Materials

Lung Cancer Data

- WES + WTS
- Normal + {Primary, CIS AIS, AAH, Dysplasia, MIA}
- Total 112 samples

Methods

Data pre-processing for variant discovery

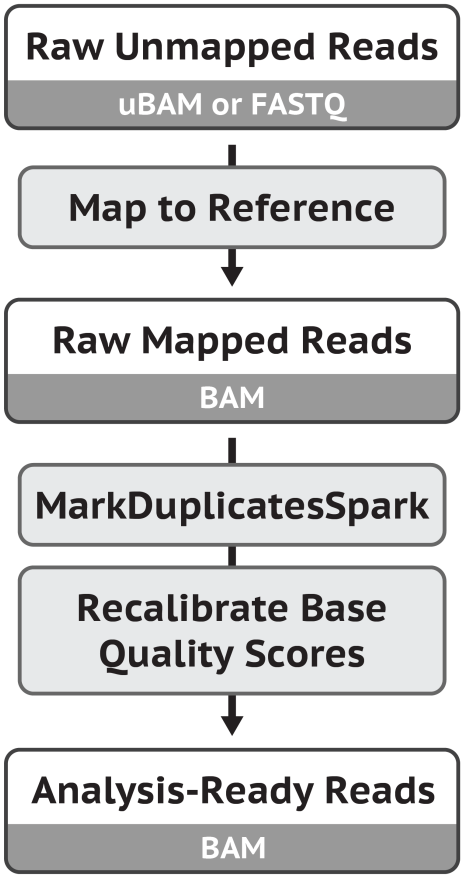

Figure: Data pre-processing for variant discovery (Van der Auwera et al., 2013; DePristo et al., 2011)

Somatic short variant discovery

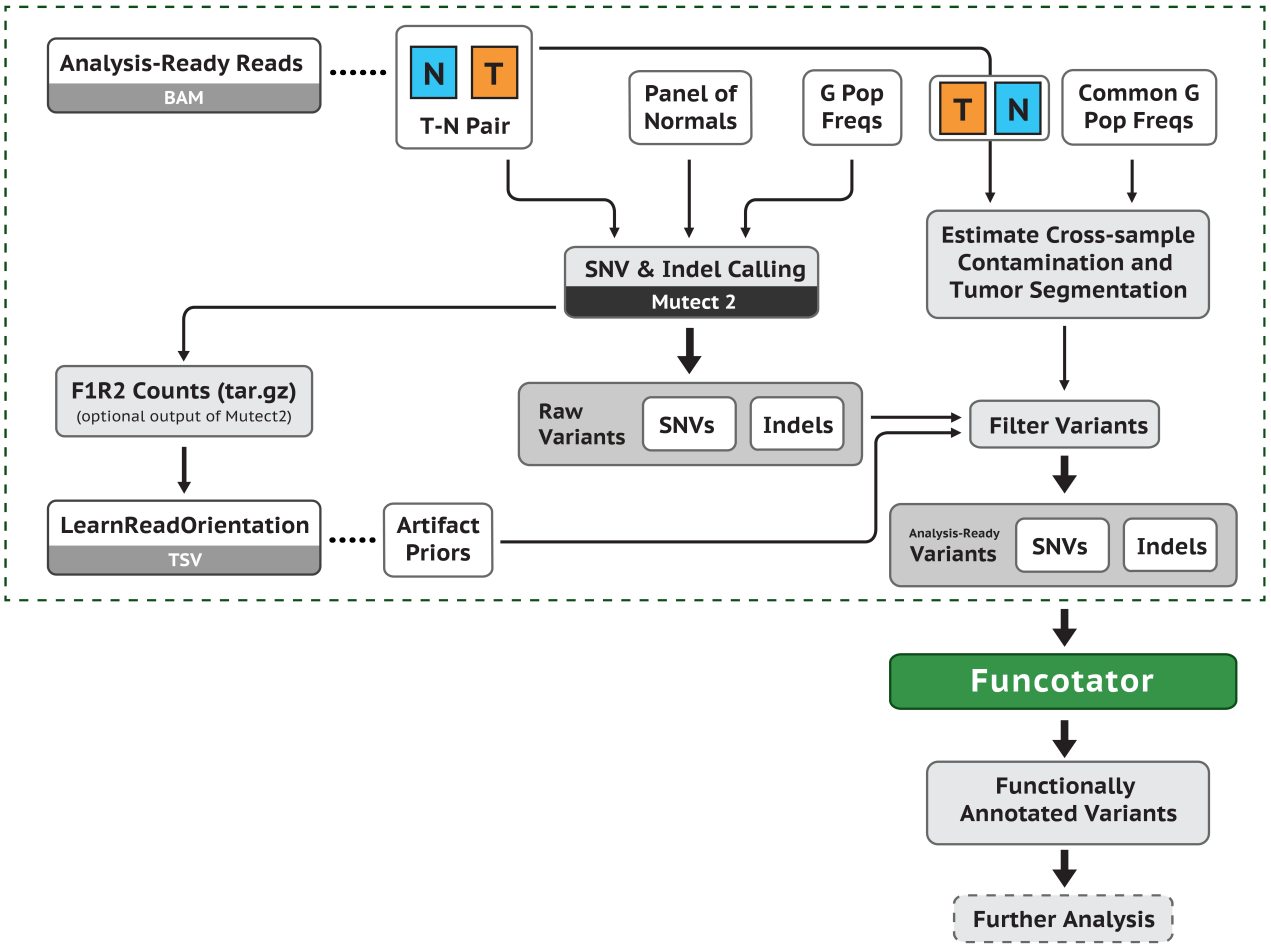

Figure: Somatic short variant (SNVs + Indels) discovery workflow (Van der Auwera et al., 2013; DePristo et al., 2011)

RNA-seq short variant discovery

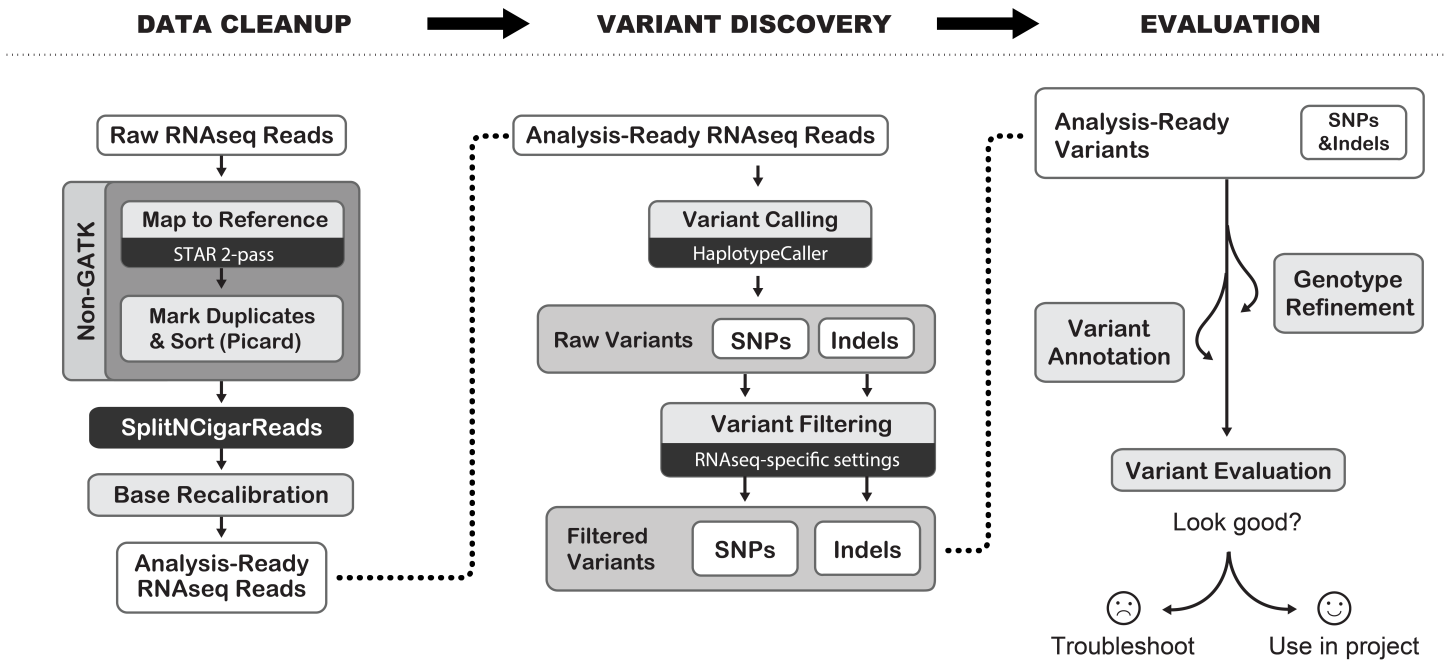

Figure: RNA-seq short variant (SNVs + Indels) discovery workflow (Van der Auwera et al., 2013; DePristo et al., 2011)

Results

FastQC

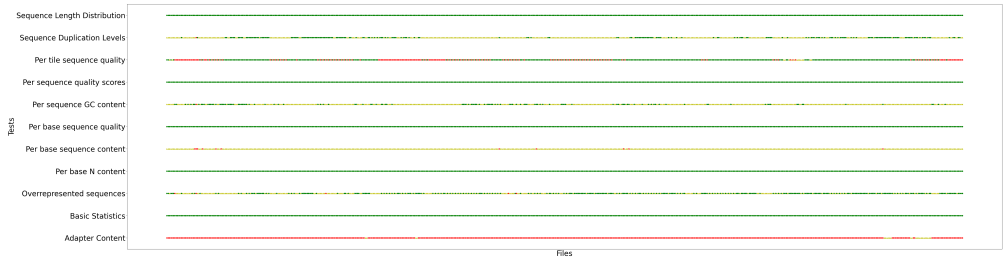

Figure: FastQC with WES data

Figure: FastQC with WTS data

$\therefore$ All PASS at *per base sequence quality*!

Proceedings

To-do Lists I

- Alignment WES with BWA
- Alignment WES with Bowtie2

References I

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.