

# Lung Cancer

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# Introduction

# Lung Cancer

# Study 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
  - from bronchus
- Diagnostic performace

# Materials

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

Materials

Cancer Types



- Carcinoma *in situ* + Adenocarcinoma *in situ*

- Atypical adenomatous hyperplasia

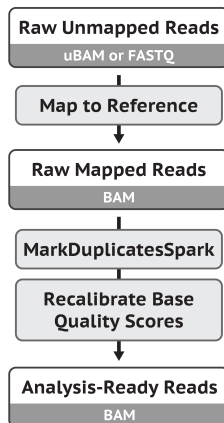
# Dysplasia



- Minimally invasive adenocarcinoma

## 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

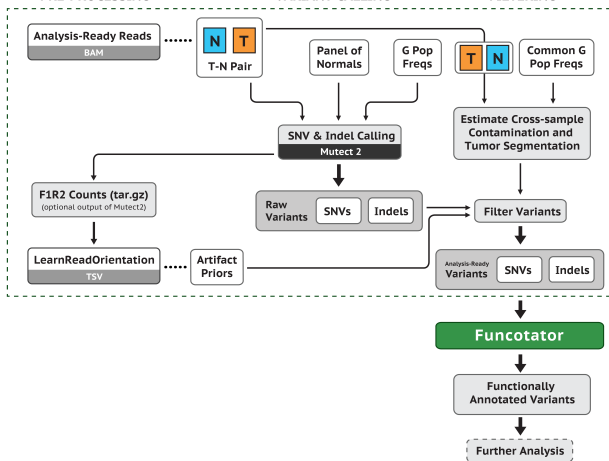


Figure: Somatic short variant (SNVs + Indels) discovery workflow (Van der Auwera et al., 2013; DePristo et al., 2011)

# RNA-seq short variant discovery

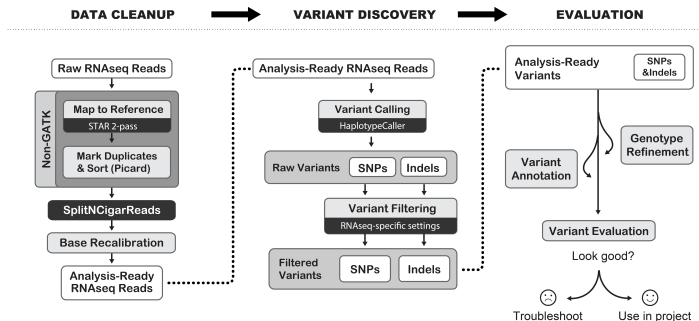


Figure: RNA-seq short variant (SNVs + Indels) discovery workflow (Van der Auwera et al., 2013; DePristo et al., 2011)



## Results

## Results

## Quality Checks

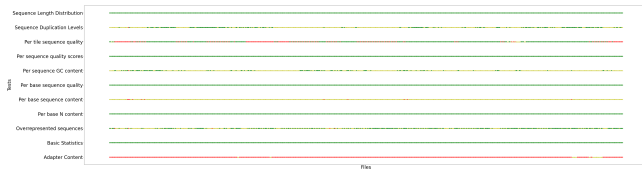


Figure: FastQC with WES data

∴ Only 33P1 has more than 3 failures: 6 FAILs.

∴ 33P1 is excluded at further analysis.

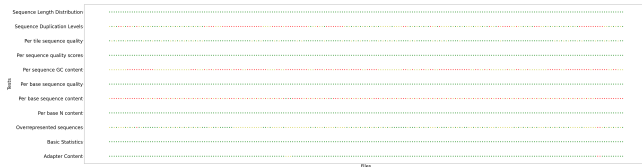


Figure: FastQC with WTS data

∴ No sample has more than 5 failures.

∴ All sample are good to analysis.

# Proceedings

# To-do Lists I

- Alignment WES with BWA
- Alignment WES with Bowtie2

- 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.