

Lung Cancer

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Overview

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Introduction

Materials

Lung Cancer Data

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

Methods

Somatic short variant discovery

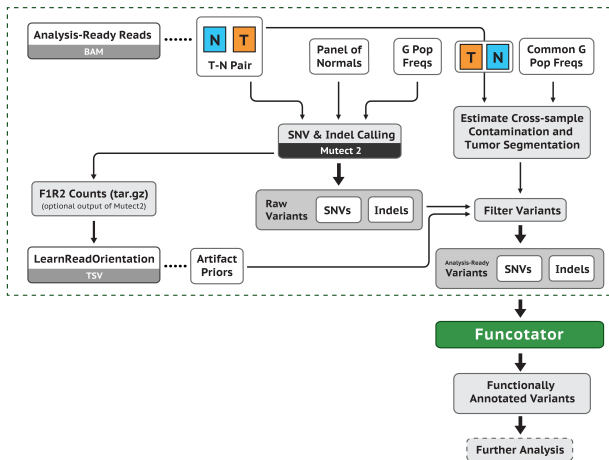


Figure: Somatic short variant (SNVs + Indels) discovery workflow (Van der Auwera et al., 2013; DePristo et al., 2011)

Somatic CNV discovery

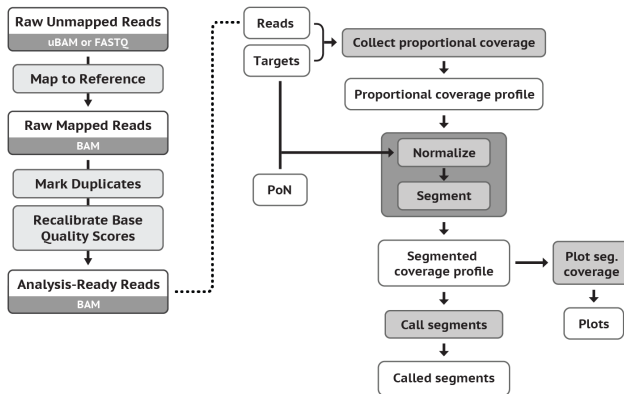


Figure: Somatic CNV (copy number variant) discovery workflow (Van der Auwera et al., 2013; DePristo et al., 2011)

Results

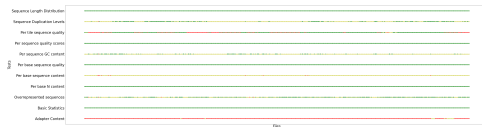


Figure: FastQC with WES data

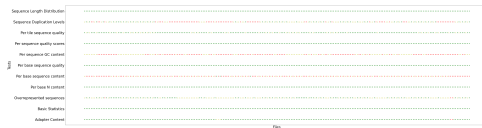


Figure: FastQC with WTS data

∴ All PASS at *per base sequence quality*!

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.