

# NGS CMC

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## Exercise until 4/7 (I will ask.)

- Find meaning of variants
  - ERBB2 p.Ile655Val
  - BARD1 R378S
  - AURKA F31I
  - KMT2C Y816\*

## Primary purpose

- Find drugable genomic alteration
- Let's list up

## My workflow

## Check whether the variants found is relevant for the cancer type

- EGFR for lung cancer
- KRAS, APC for colon cancer
- If in doubt, look at histological diagnosis and the unusual variants

## Look at the variants called

- Unusual variants (possible false positive)
- Germline polymorphism
  - Allele frequency
  - ClinVar <https://www.ncbi.nlm.nih.gov/clinvar/> (<https://www.ncbi.nlm.nih.gov/clinvar/>)
  - <https://gnomad.broadinstitute.org/> (<https://gnomad.broadinstitute.org/>)
- Alignment error
  - Frameshift variant
  - IGV
- Splicing site variants
  - VUS
  - ClinVar
- Low read count fusion
  - Less than 1000 readcount
  - Primer-dimer
  - Other methods IHC, FISH

## Check IGV

## Check Whitelist

- Check QC
  - Pass is not enough
    - Total mapped RNA read
    - > 500,000 is not enough
    - MAPD for CNV
- Check filter out variants by sorting

## Check BRCA1/2 for breast, ovary, pancreas and prostate

- Filter out variants by sorting
- myBRCAvariant\_filter
- <https://www.ncbi.nlm.nih.gov/clinvar/> (<https://www.ncbi.nlm.nih.gov/clinvar/>)
- Evaluate the variants <https://www.sciencedirect.com/science/article/pii/S0002929716300349?via%3Dihub> (<https://www.sciencedirect.com/science/article/pii/S0002929716300349?via%3Dihub>)

## Check annotations

- Examples
  - EGFR Exon20 deletion
  - <http://varnomen.hgvs.org/> (<http://varnomen.hgvs.org/>)

## Interpretation

- <http://10.10.56.106:3729/> (<http://10.10.56.106:3729/>)
- <https://www.cbioportal.org/> (<https://www.cbioportal.org/>)
- <https://civicdb.org/home> (<https://civicdb.org/home>)
- <https://www.mycancergenome.org/> (<https://www.mycancergenome.org/>)

## Note edit

## What to do

- Memo on printed NGS management system
- Fill in interpretation
- TAT is important

## Server address

- IR <https://10.10.55.96/ir/login.html> (<https://10.10.55.96/ir/login.html>)
- OR <http://10.10.55.96:8088/#/> (<http://10.10.55.96:8088/#/>)
- NGS management system <http://160.1.17.79/#/login> (<http://160.1.17.79/#/login>)