# Machine Learning for Cancer Genomics

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March 23, 2024

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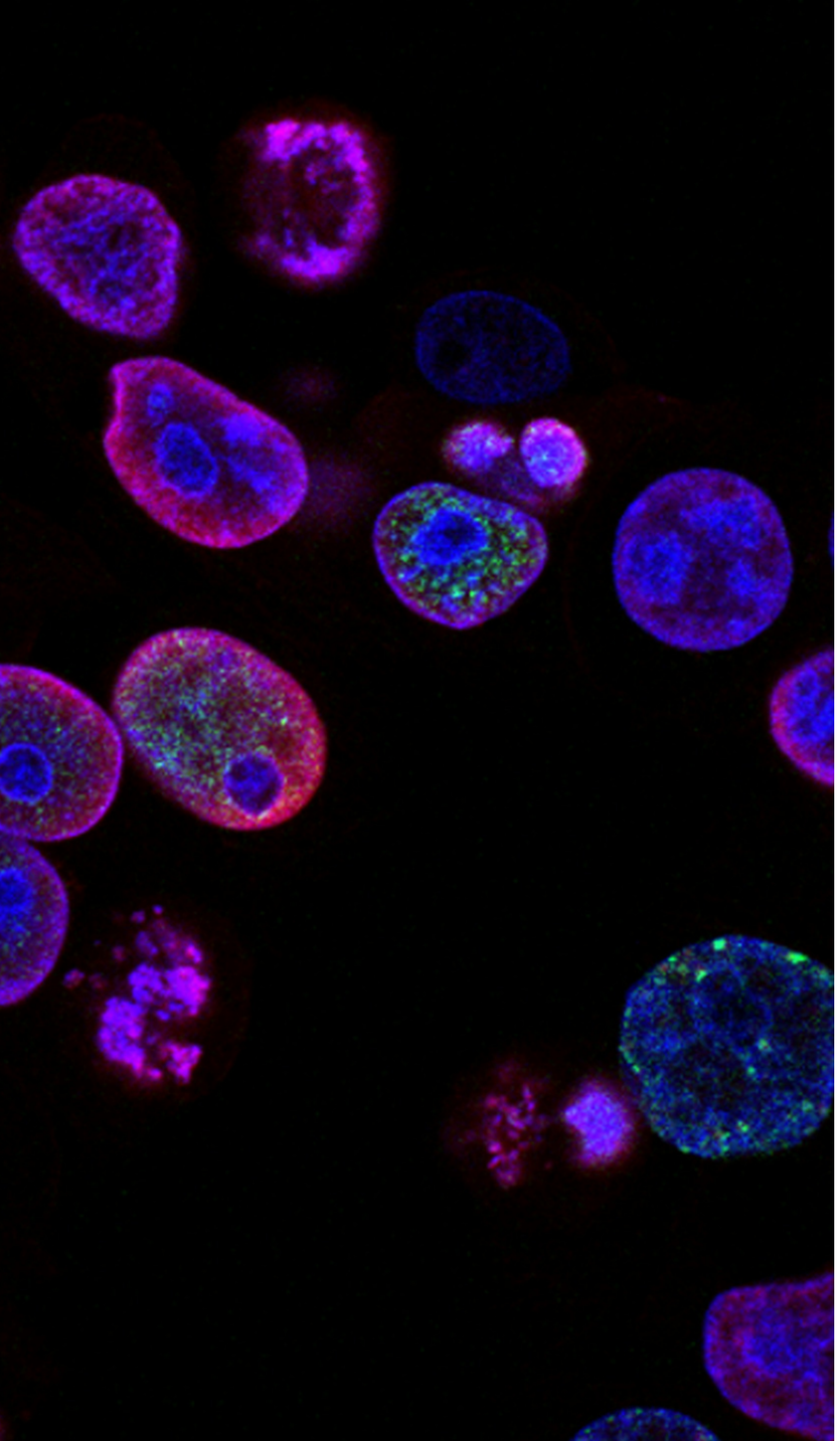
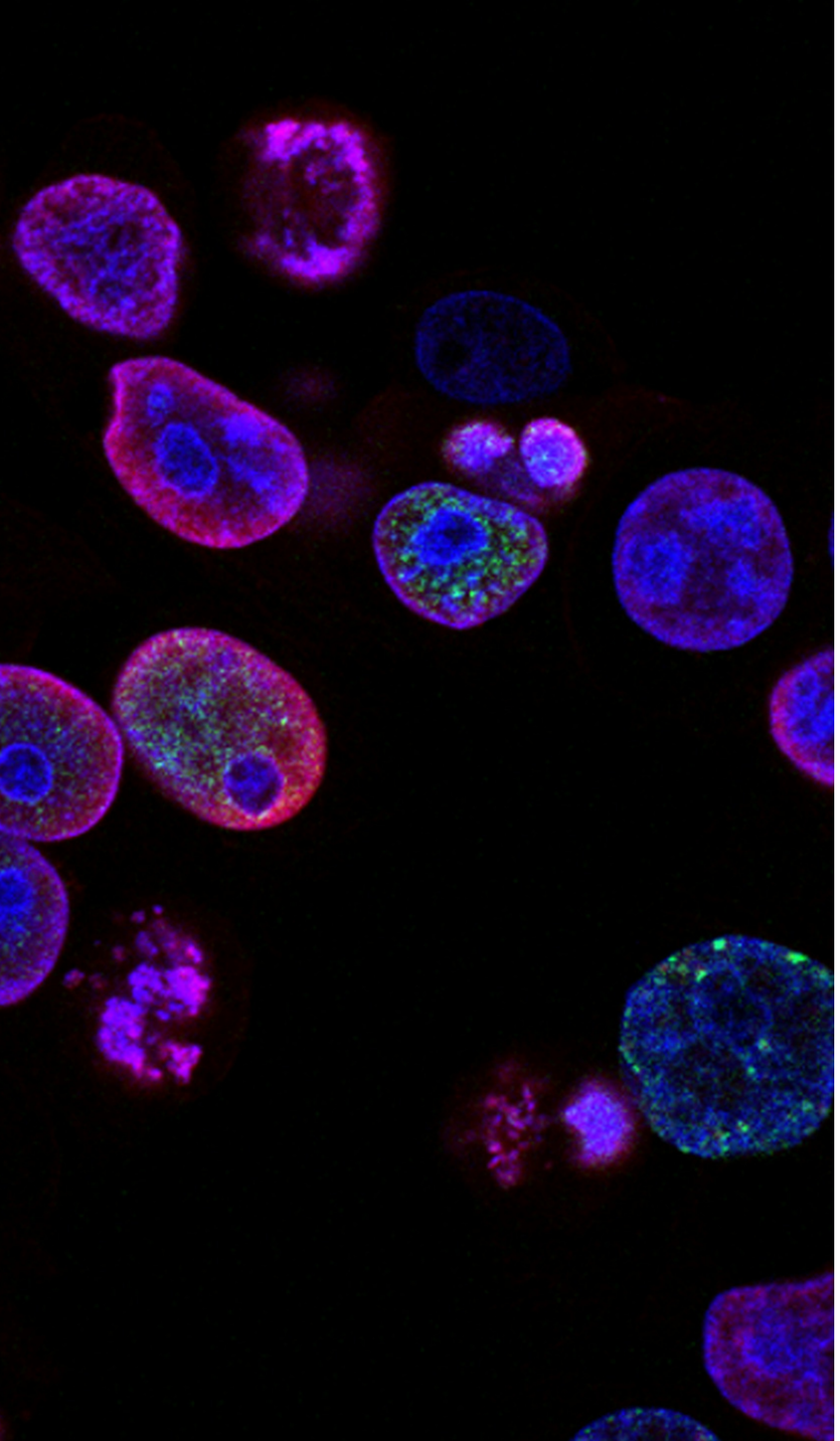


# Agenda

Part 1: Brief overview of the field

Part 2: My research - cancer evolution

Part 3: A day in the life of a PhD student



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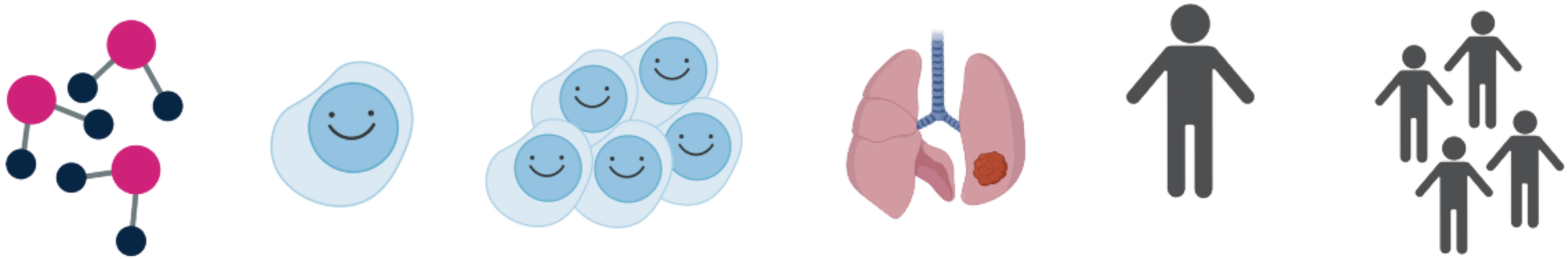
## Computational biology

## ML for health

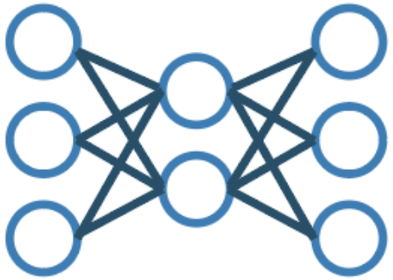
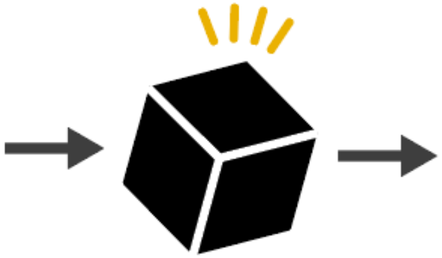


# Challenges in biomedical ML applications

- Data size
- Labels are expensive
- Missing “ground truth”
- Generalization
- Interpretability and causal inference

# Two common modelling paradigms

# Two common modelling paradigms

| | Input | Training loop | Final product |
|---|---|---|---|
| **Deep learning for prediction** |  Data |  Update weights |  Black box predictor |

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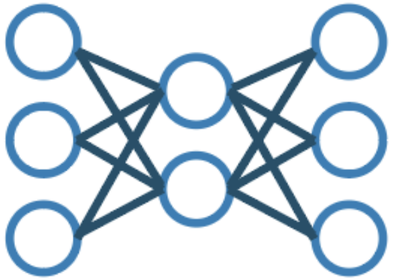
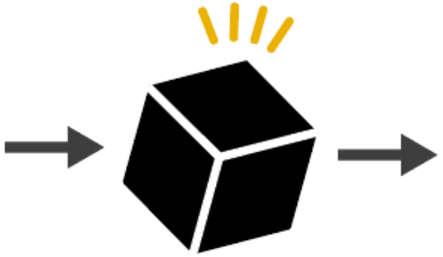
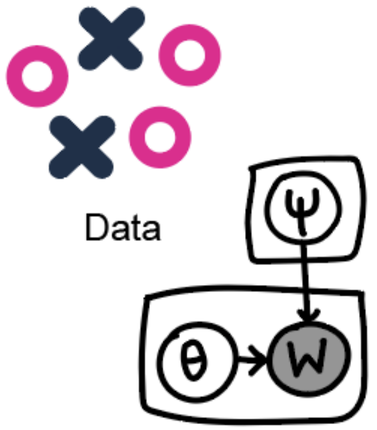
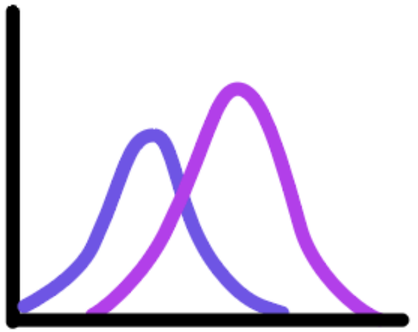
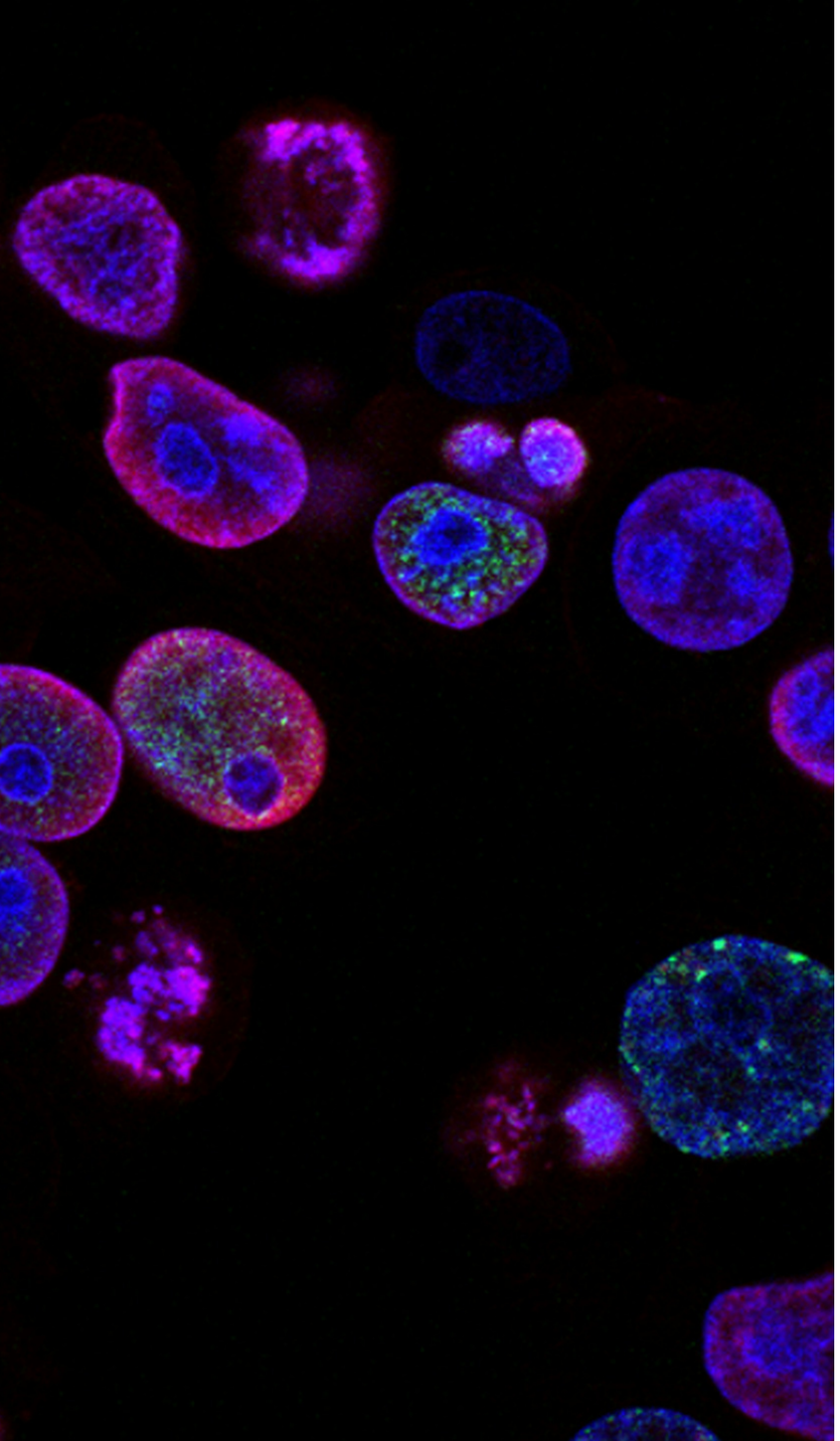
| **Deep learning for prediction** |  Data |  Update weights |  Black box predictor |
| **Bayeisan inference** |   Probabilistic model | $\hat{\theta}_i = U(\hat{\theta}_{i-1})$ Update parameter estimates |  Posterior distributions |



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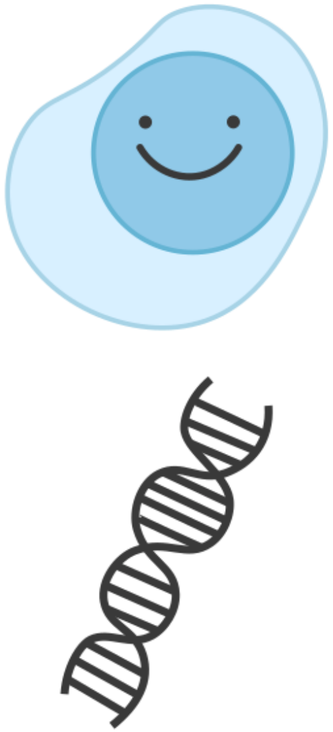
# DNA = set of instructions which tell a cell what to do



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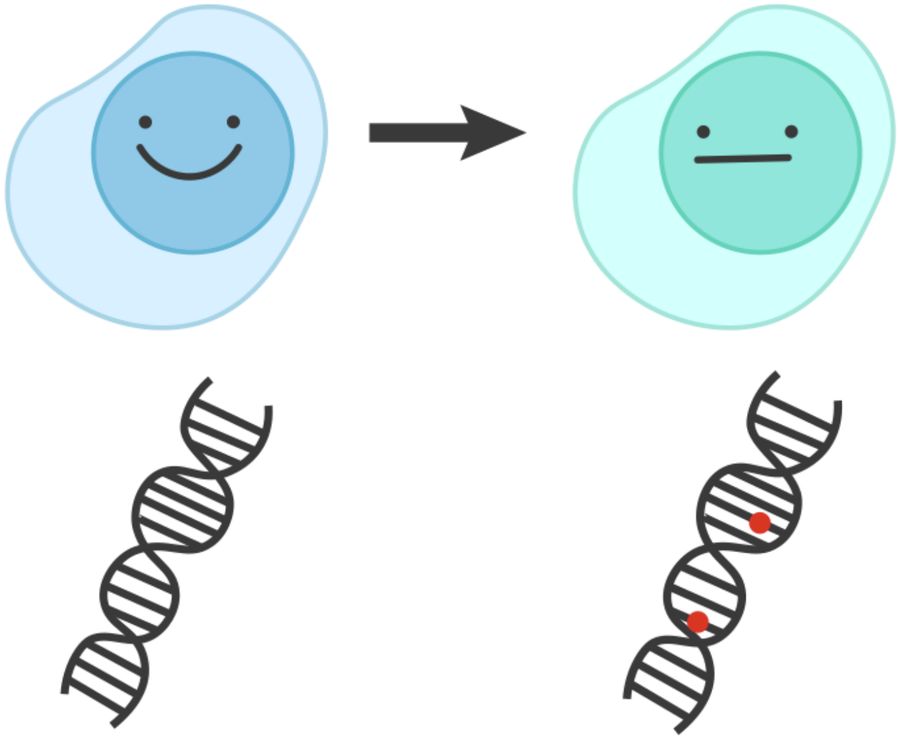

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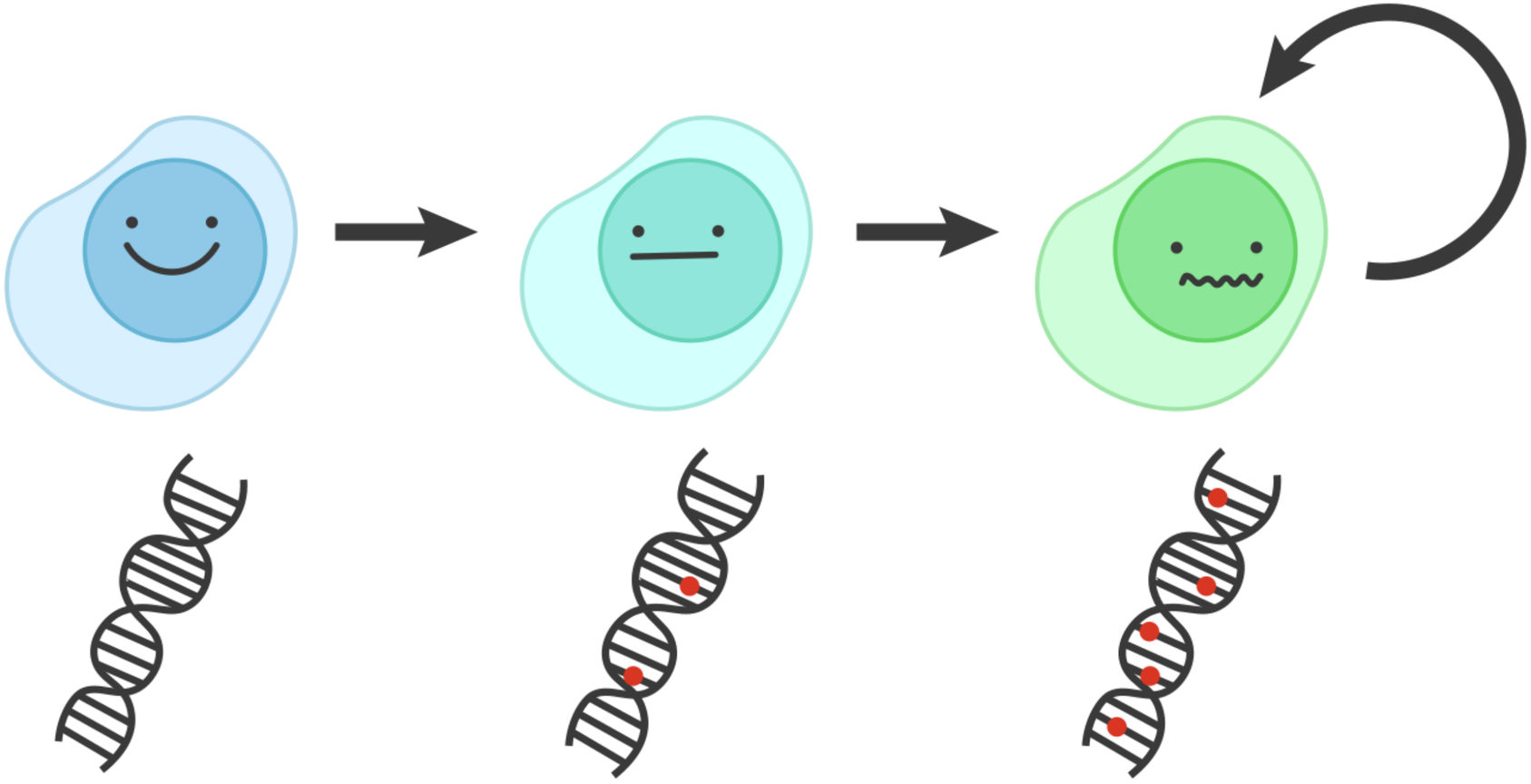
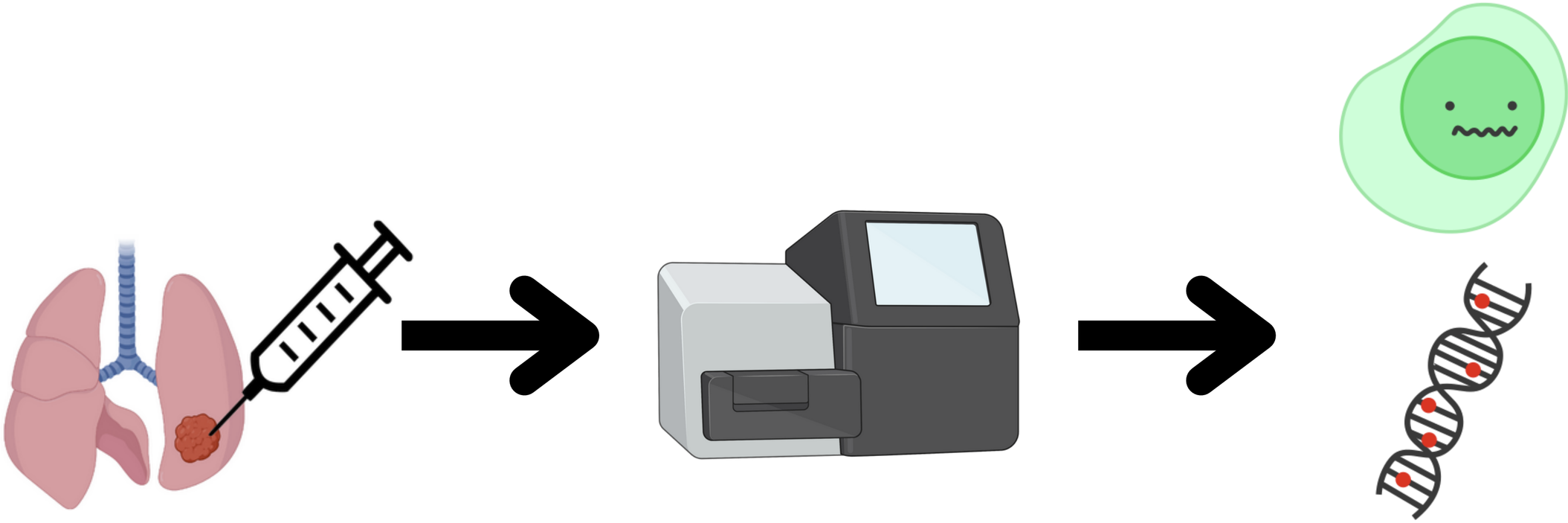


# With whole genome sequencing, get a list of mutations in a sample



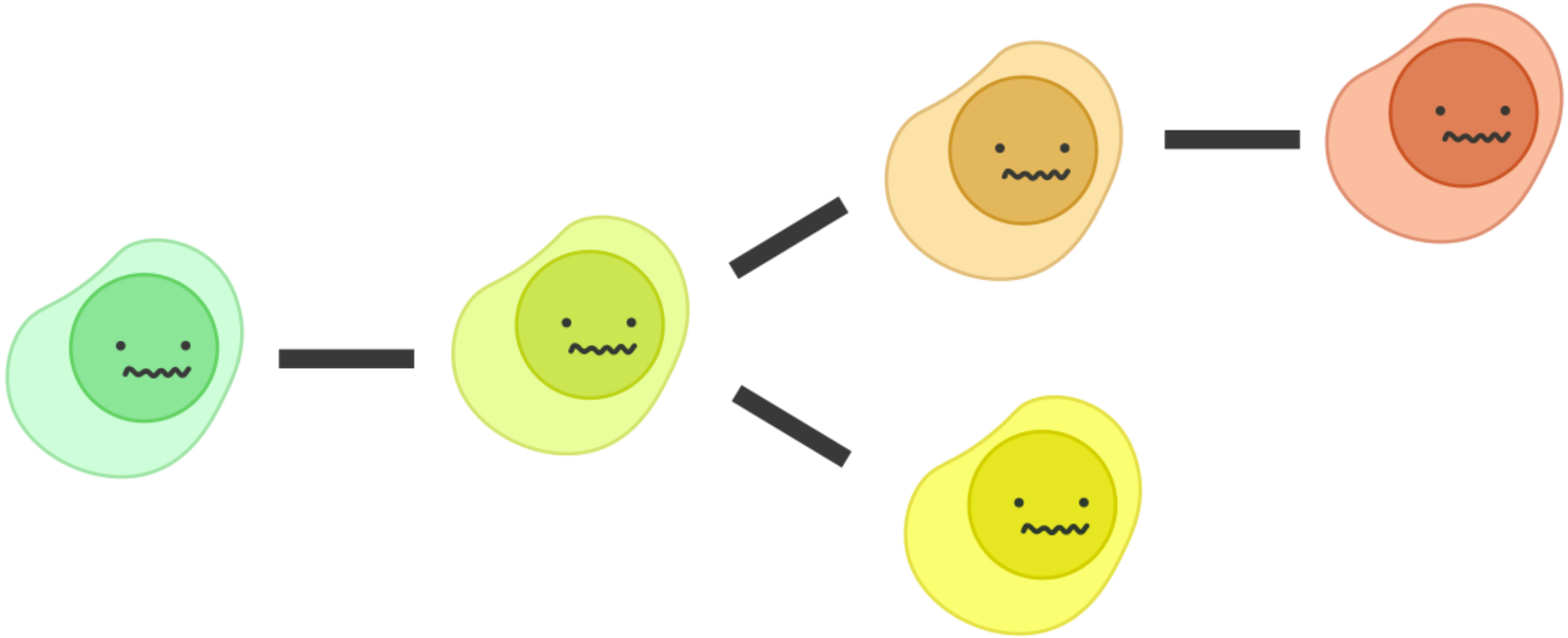
Sample

Sequence DNA

Identify mutations

# A tumour sample contains a mixture of cells!

# Cells in a tumour are all related



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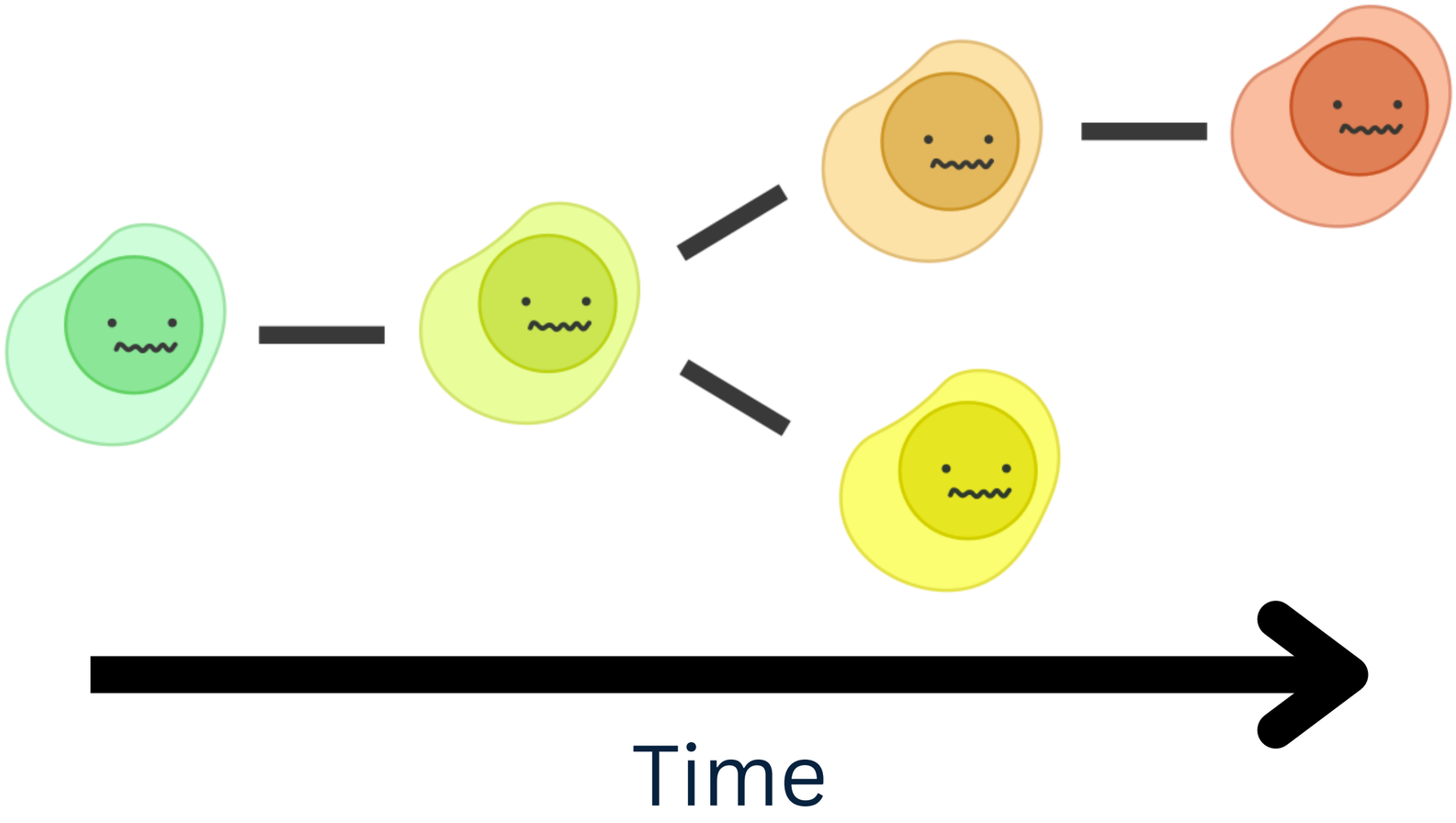
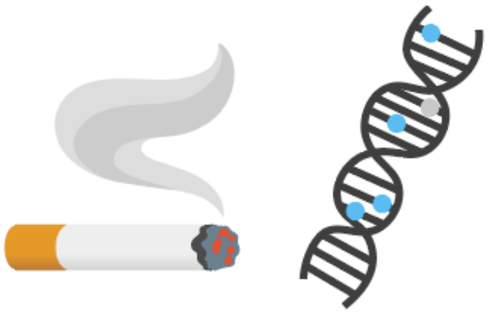
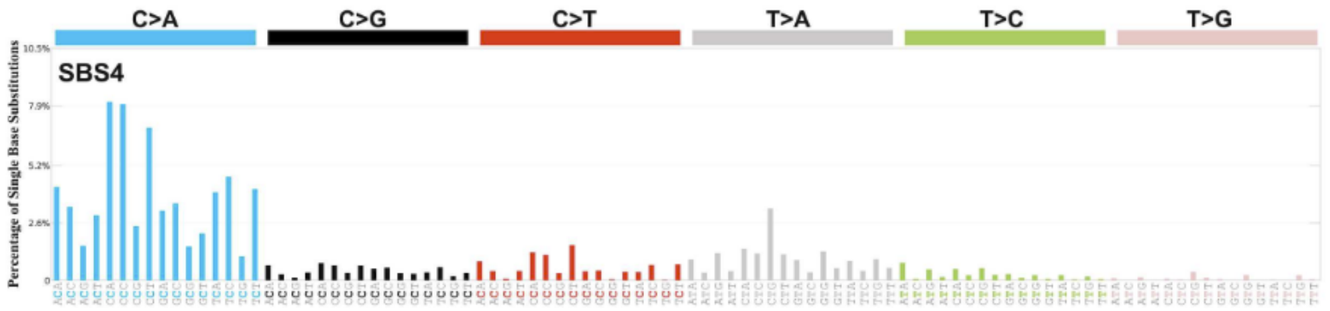
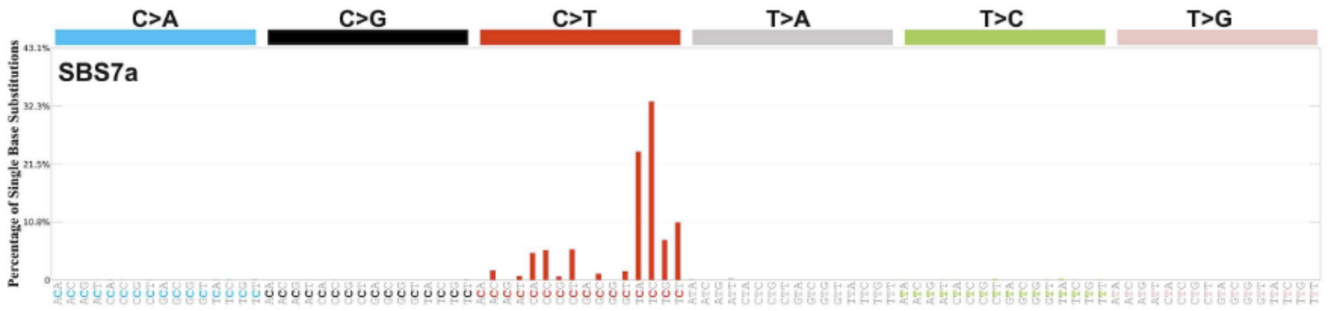


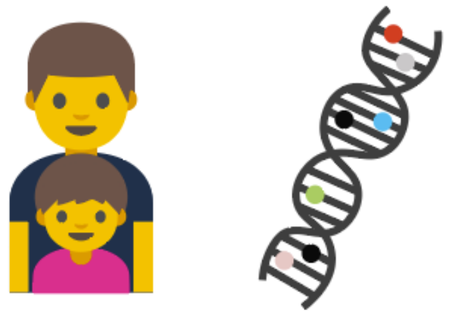

Mutations act as a “fossil record” of everything that happened to the tumour over its development.

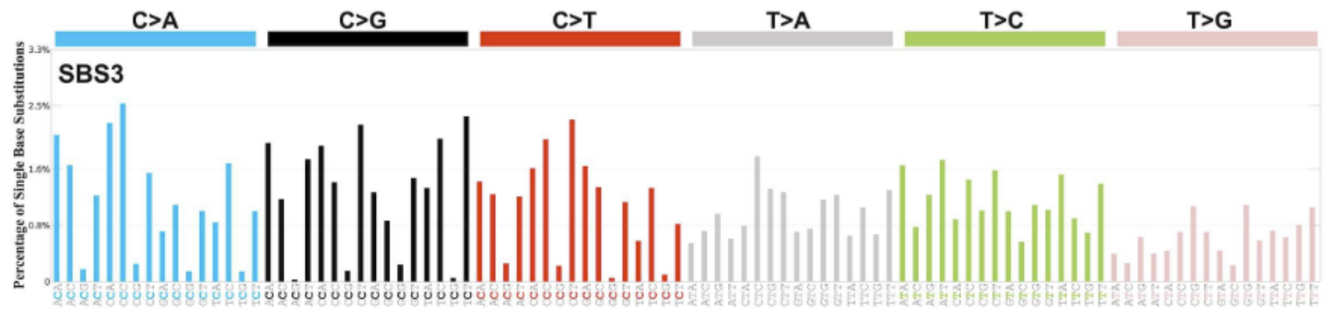
# Mutagens and DNA repair defects can cause distinct patterns of mutations



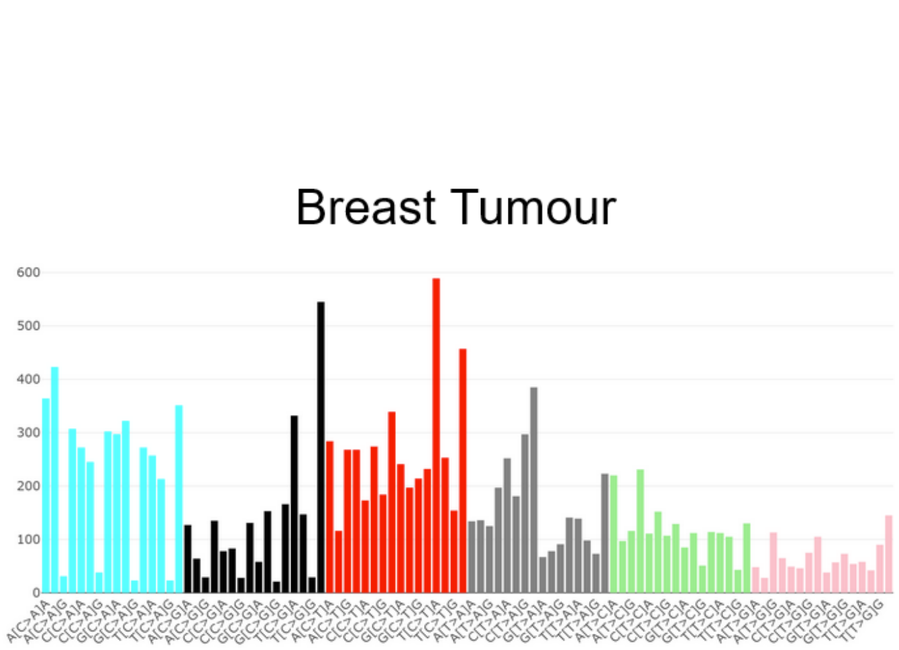




# Mutational signature analysis = identify these patterns

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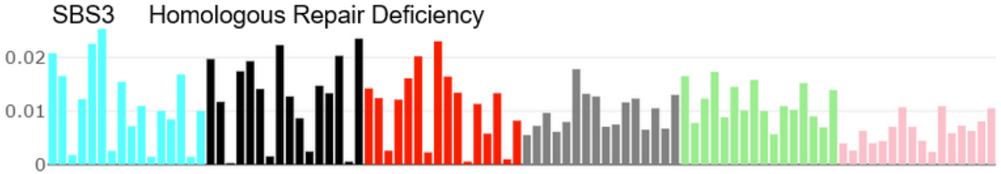
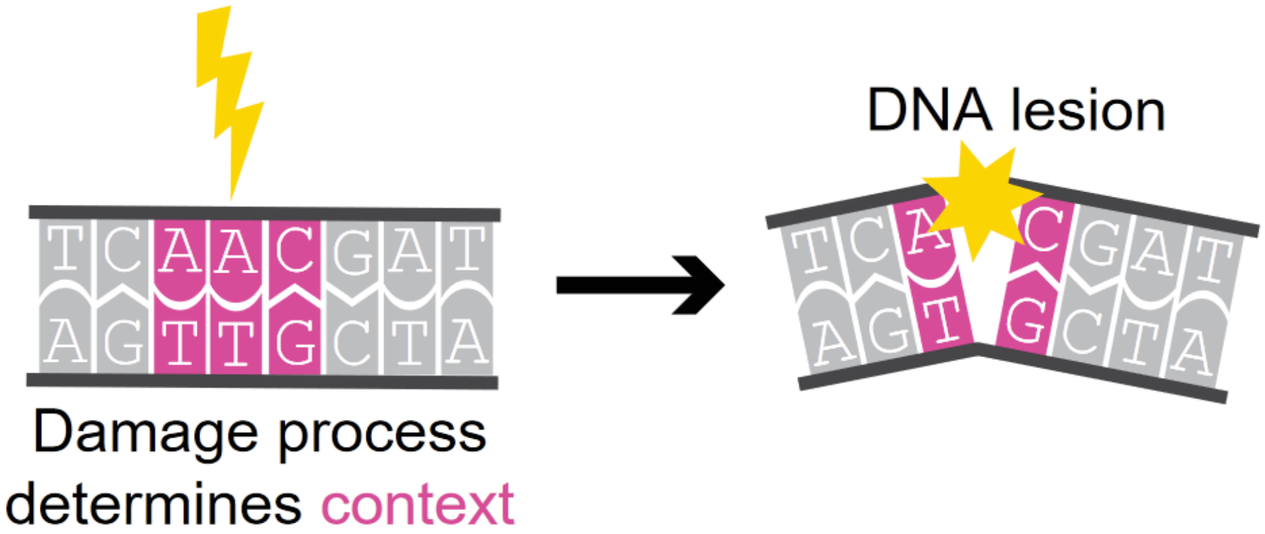





Other

# Problem: current standard model mixes the effects of damage and misrepair processes



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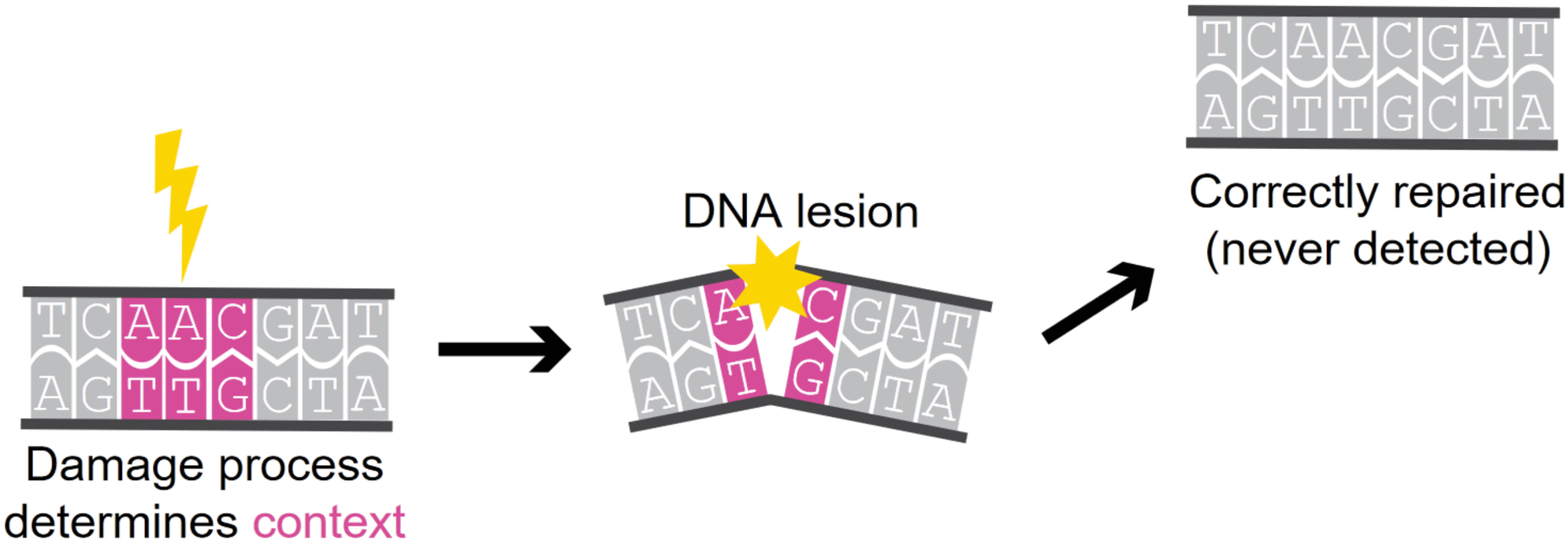

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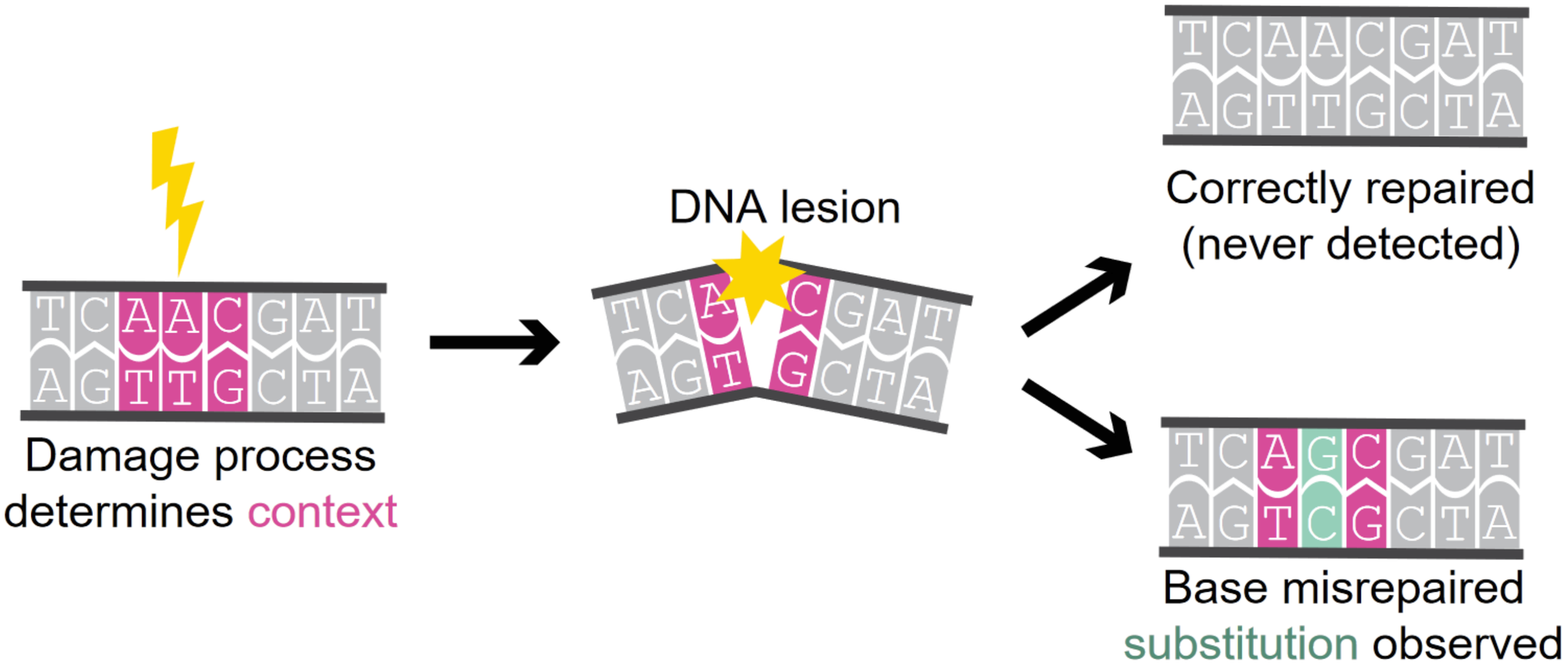
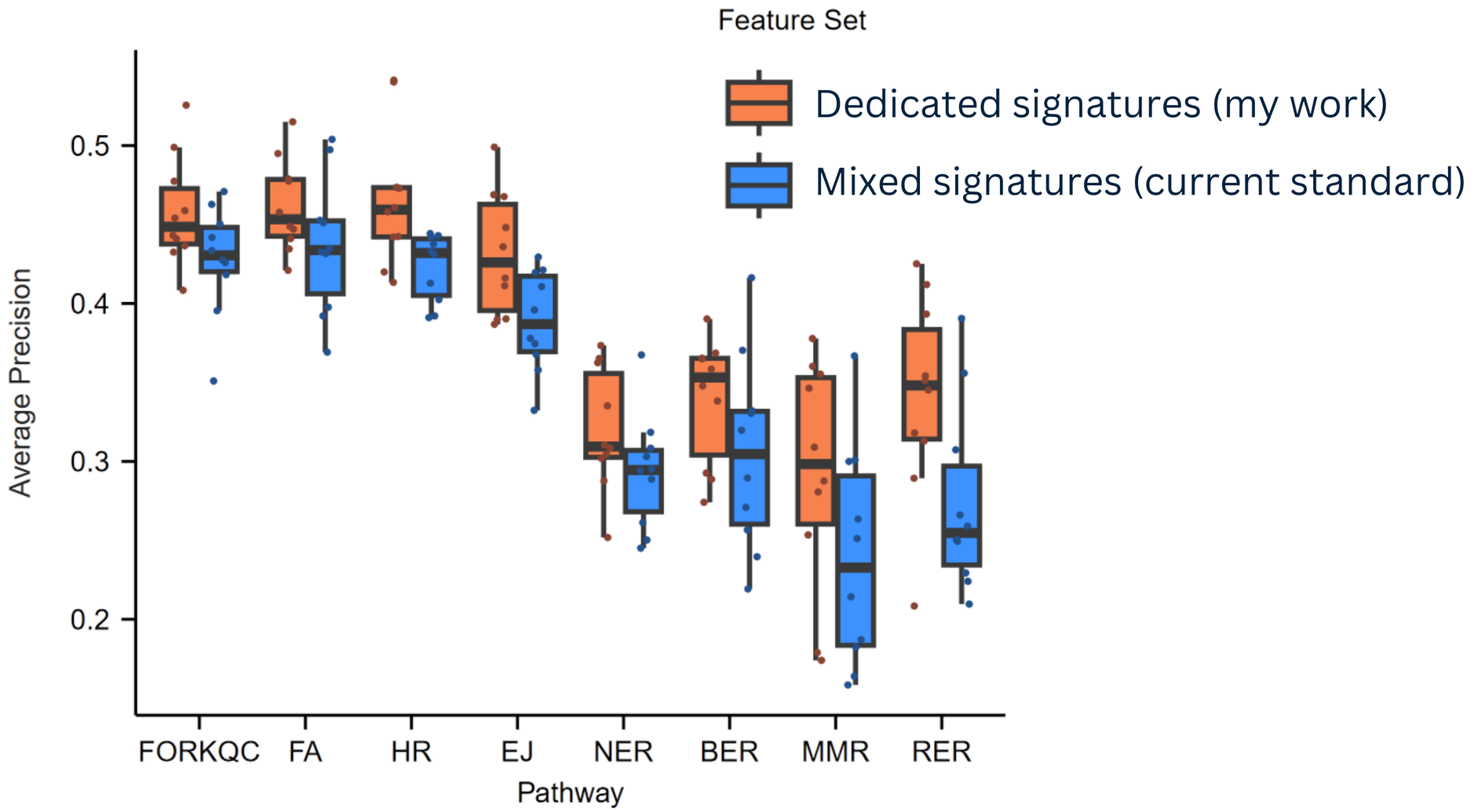
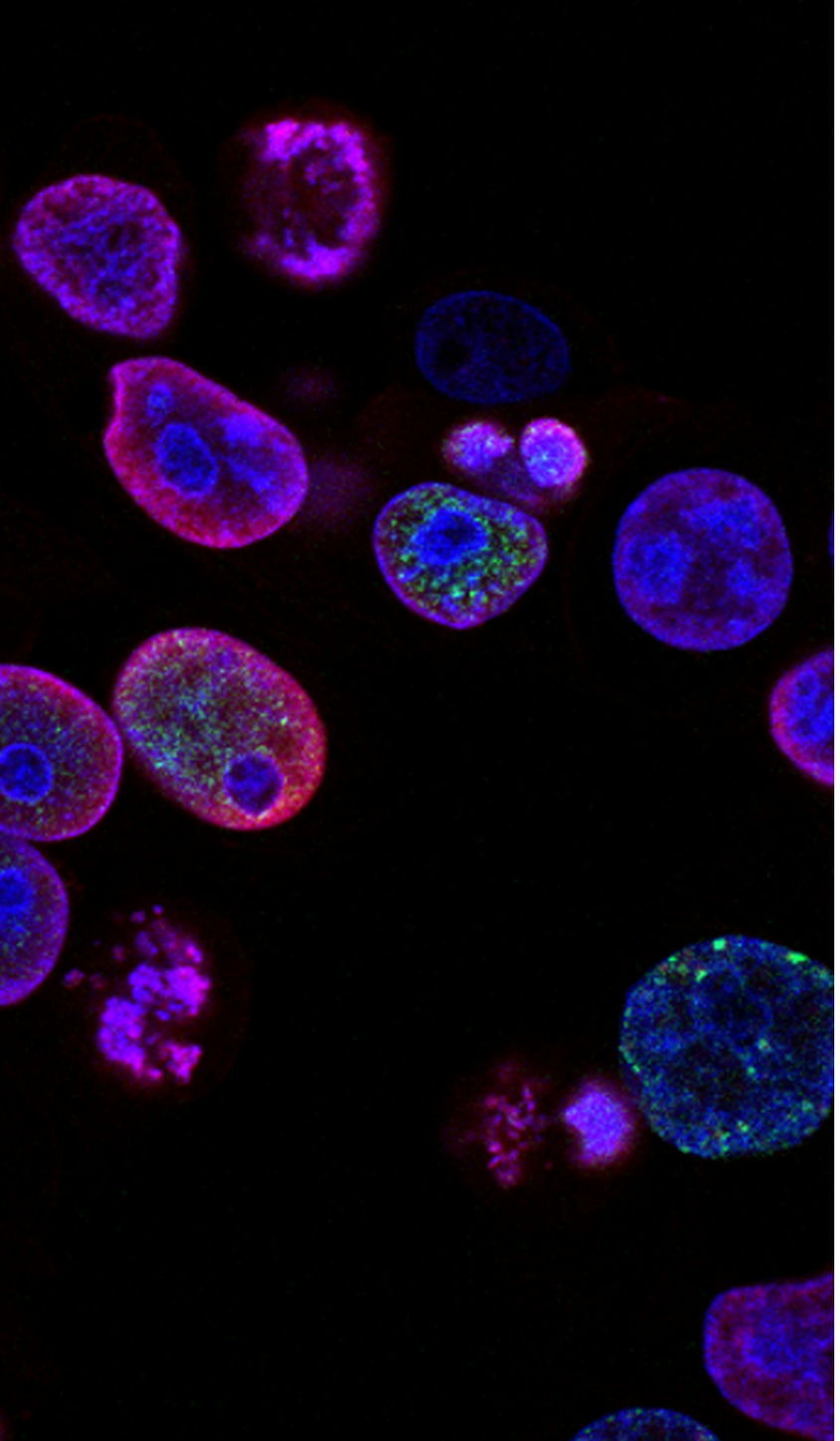


# Having dedicated damage and repair signatures helps predict repair deficiencies in cancer





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## PhD life

The good: you get to work with amazing people, on problems you think are cool.

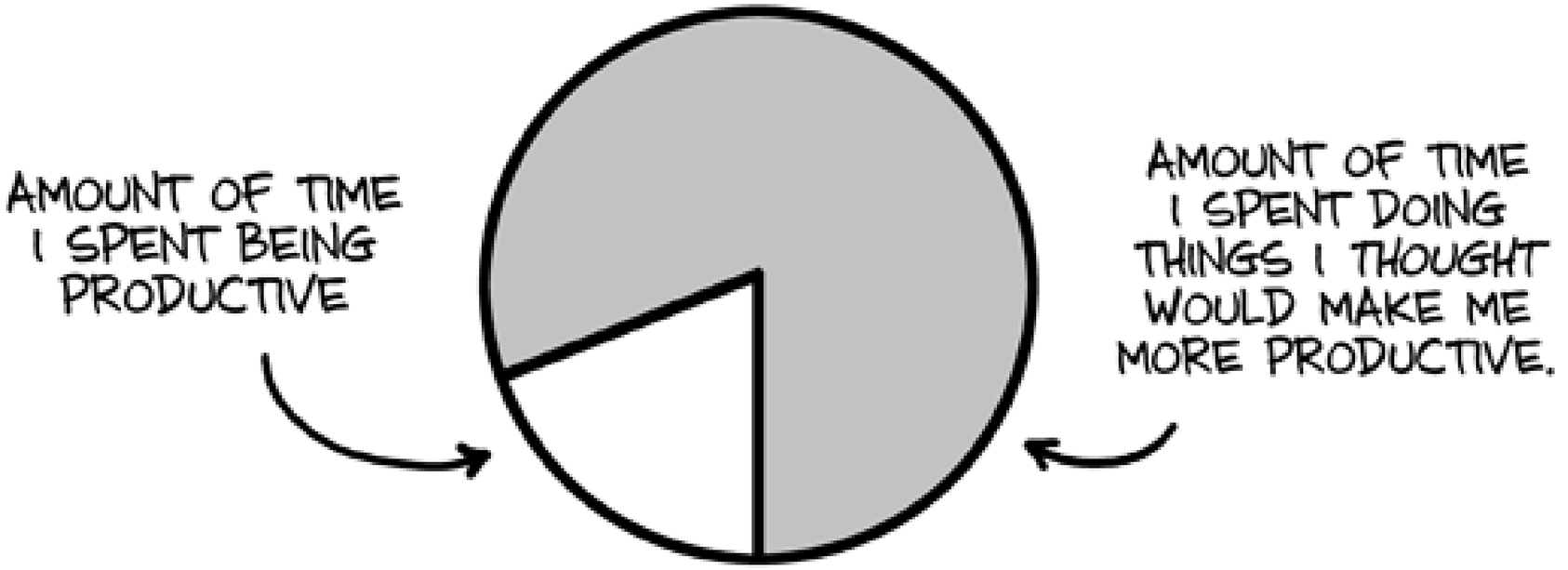
The bad: It can feel like a group project where you're the only one who cares if you get a good grade

The ugly: it takes 4+ years

# How I spend my time

- A **lot** of reading papers
- Writing code, doing data analysis
- Making plots & figures
- Writing results and scholarship applications
- Talking to other scientists

# HOW MY WEEK WENT:

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## Things you can do now to prepare for grad school

- Take a project-based class to get good at working independently
- Read papers
- Attend talks (ex. monthly meeting of torbug.org)
- Join a research lab

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