

# Models for Microsatellite Mutation

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# Microsatellites

- Repeats of a short motif, e.g. AT repeated 6 times:

A T A T A T A T A T A T

- Think of microsatellites as repeat units:

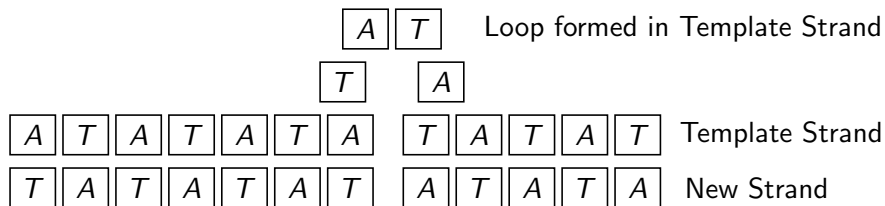
AT AT AT AT AT AT

- Highly polymorphic.
- Abundant in eukaryote genomes.

# Slipped-strand mispairing

## Contraction

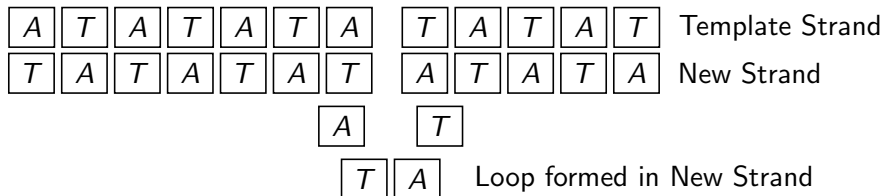
During replication, a loop may form in the template strand leading to a decrease in the number of repeats in the new strand.



# Slipped-strand mispairing

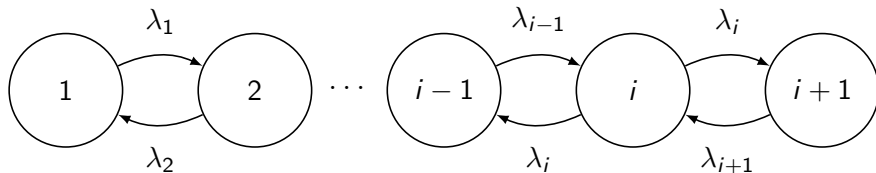
## Expansion

Alternatively, a loop may form in the new strand, leading to an increase in repeat number relative to the template.



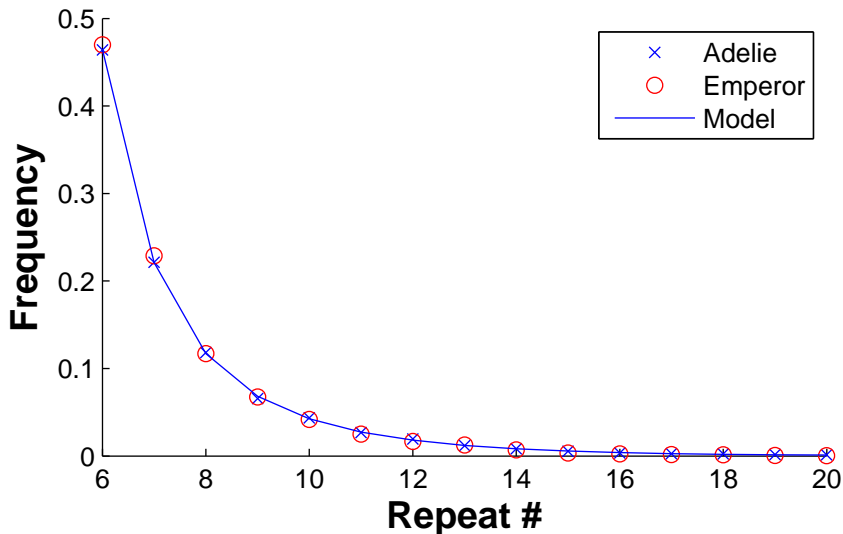
# Models for slipped-strand mispairing

- e.g. a symmetric random walk:



- The main factors accounted for are:
  - Length dependence of mutation rate.
  - Bias towards contraction or expansion.
  - Size of the mutation events.

# Adelie vs Emperor AC distribution



# Point Mutation

- Microsatellites also susceptible to point mutations.

AT AT AT AC AT AT

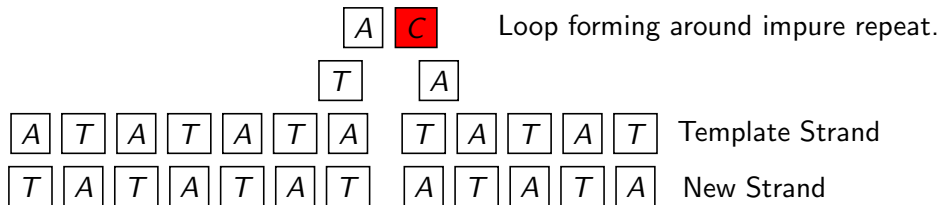
- How to deal with this?

AT AT AT

AT AT

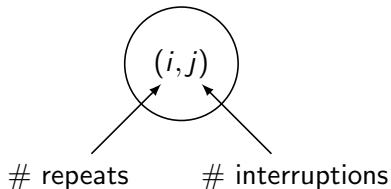
# Point Mutation

- These models lose useful information, and may invalidate IID assumption.





- We move up a dimension in the state space.

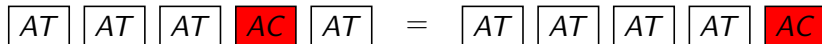


$AT$   $AT$   $AT$   $AC$   $AT$   $AT$

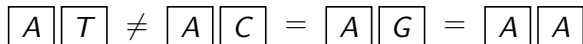
$= (6, 1)$

# Key Assumptions

- Effect of impurity is independent of location.



- Each base pair is either 'correct' or 'incorrect'.



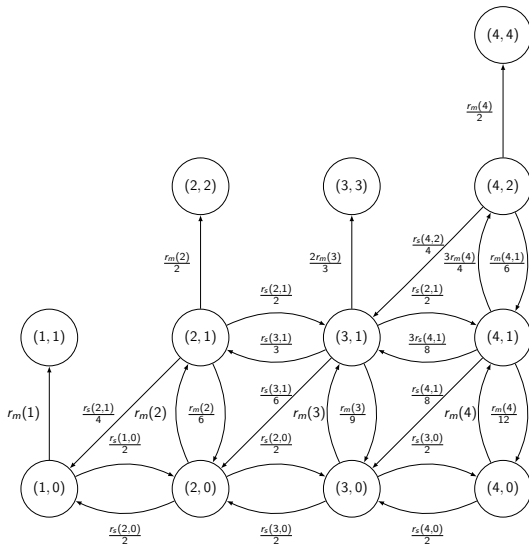
# Extra Assumptions

- A repeat *unit* is either pure or impure - binary.

$$\boxed{AT} \neq \boxed{AX} = \boxed{YT} = \boxed{YX}$$

- A sequence which is more than half impure is no longer a microsatellite.
- Slippage events of length 1 only.

# Our Model



## Supervisors

- Dr Małgorzata O'Reilly
  - Dr Barbara Holland
- 
- Dr Bennet McComish