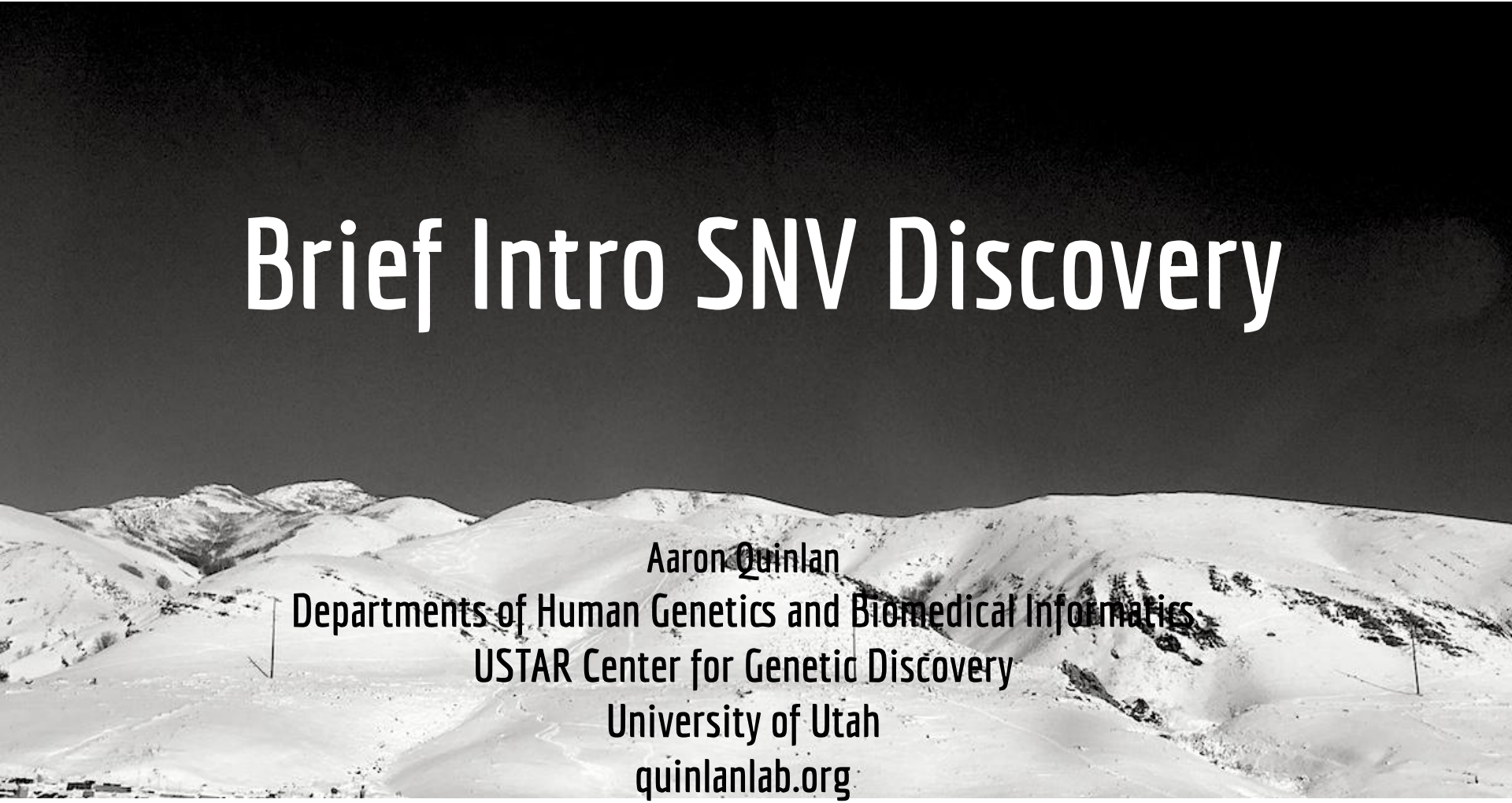
# Brief Intro SNV Discovery



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University of Utah
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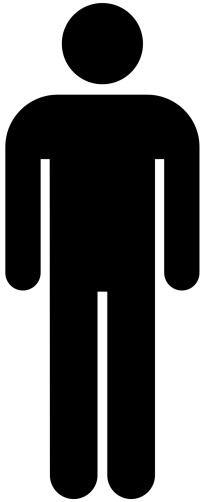
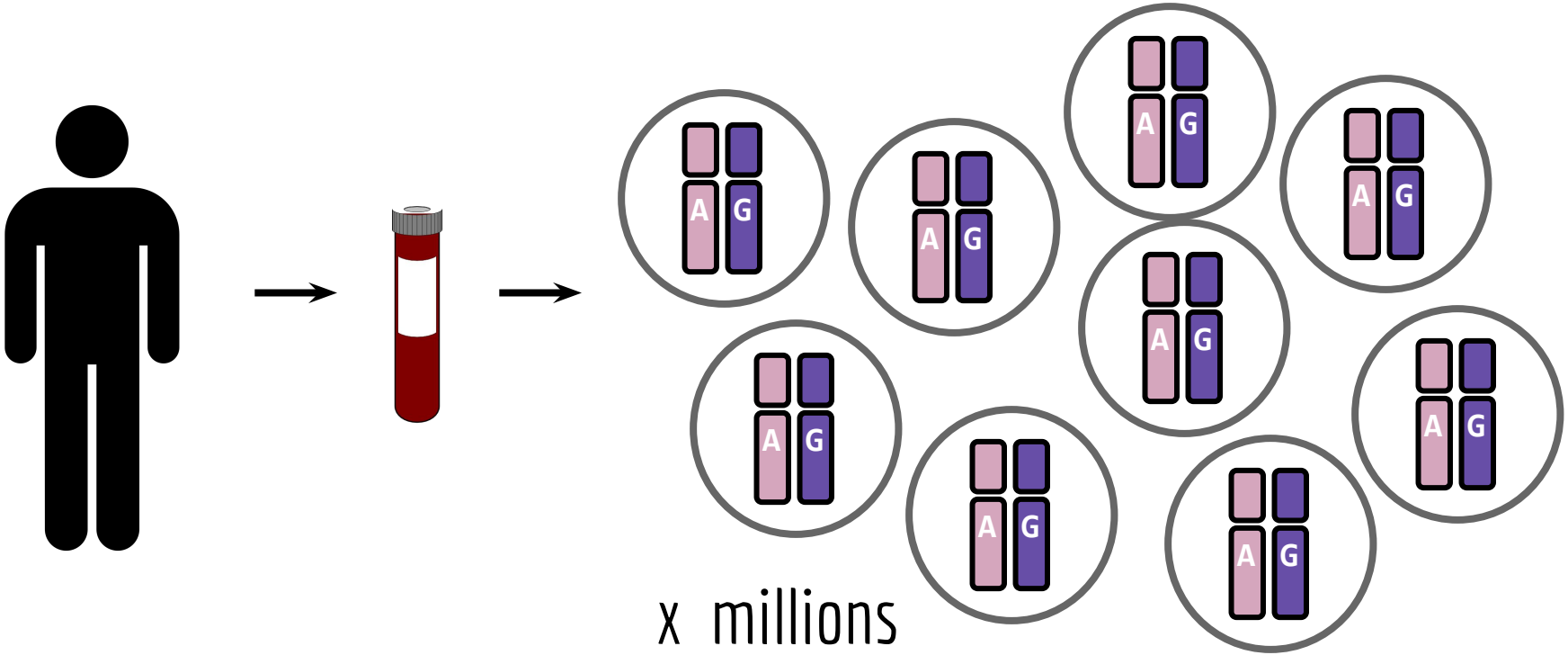
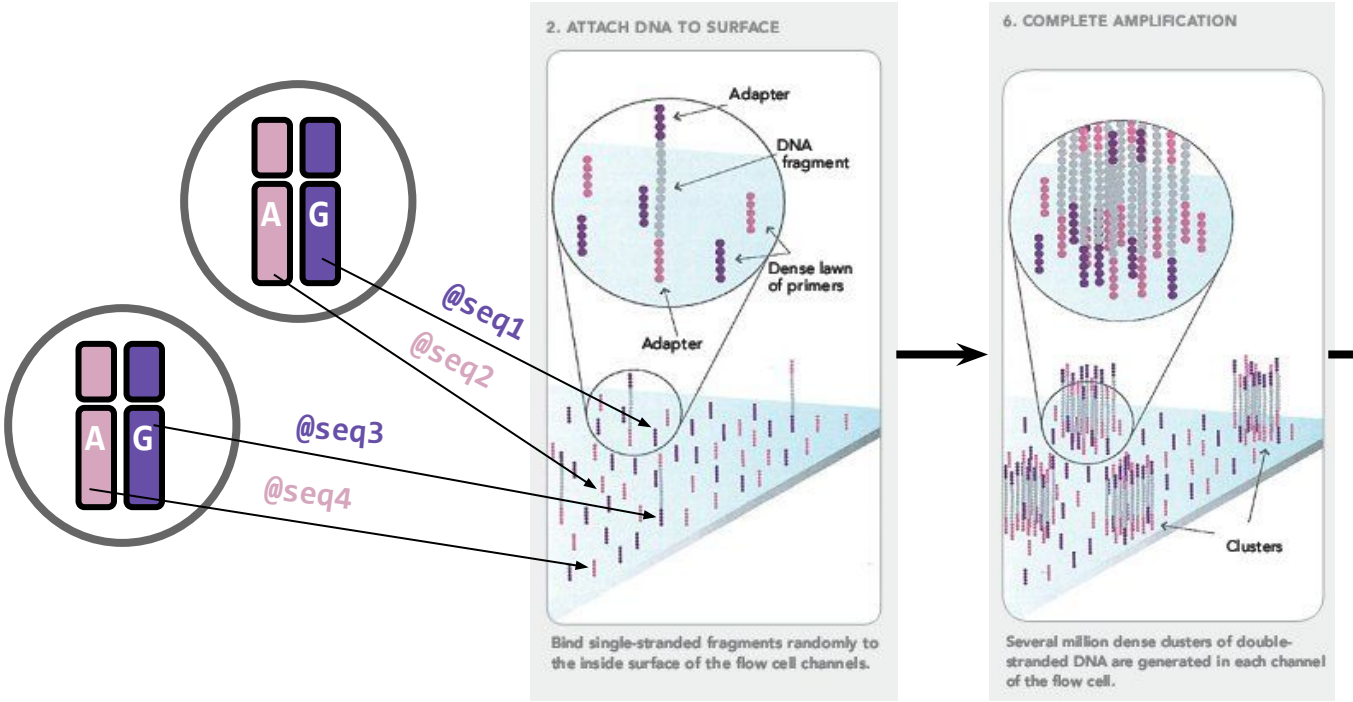
Goal: find all inherited variants in an individual's diploid genome.



# Find inherited genetic variation by sequencing DNA from millions of cells



# Each DNA cluster is amplified from a single strand from a single haploid chromosome from a single cell.




```
@seq1
ACCTTCGAACGGCGGGGGGTTACAA
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!''*((((***+))%%%++).1***
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TGGAACCGAACGGCCCCGGTTACAT
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```
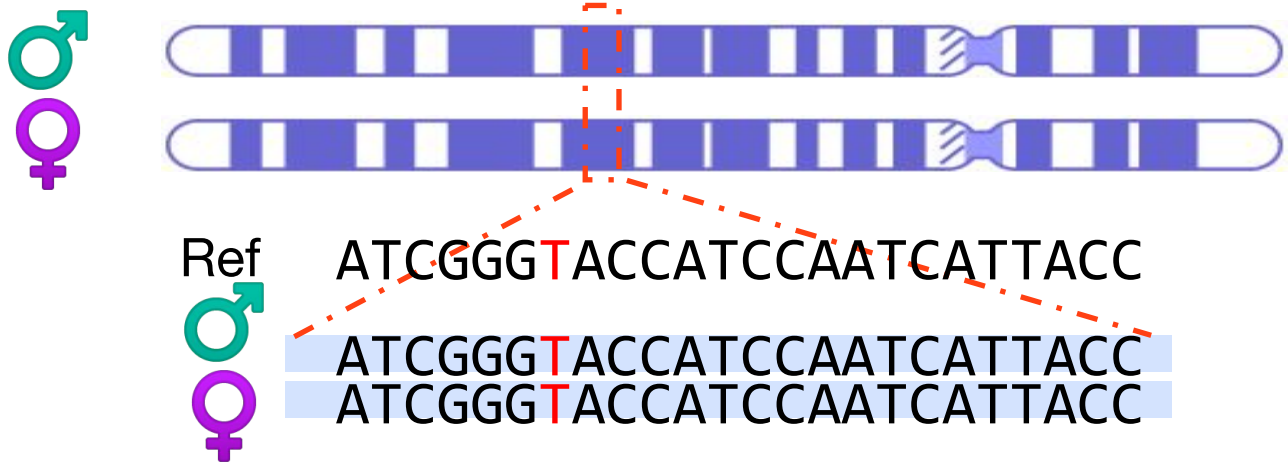

# Scenario 1: An individual is homozygous for the "reference" allele.



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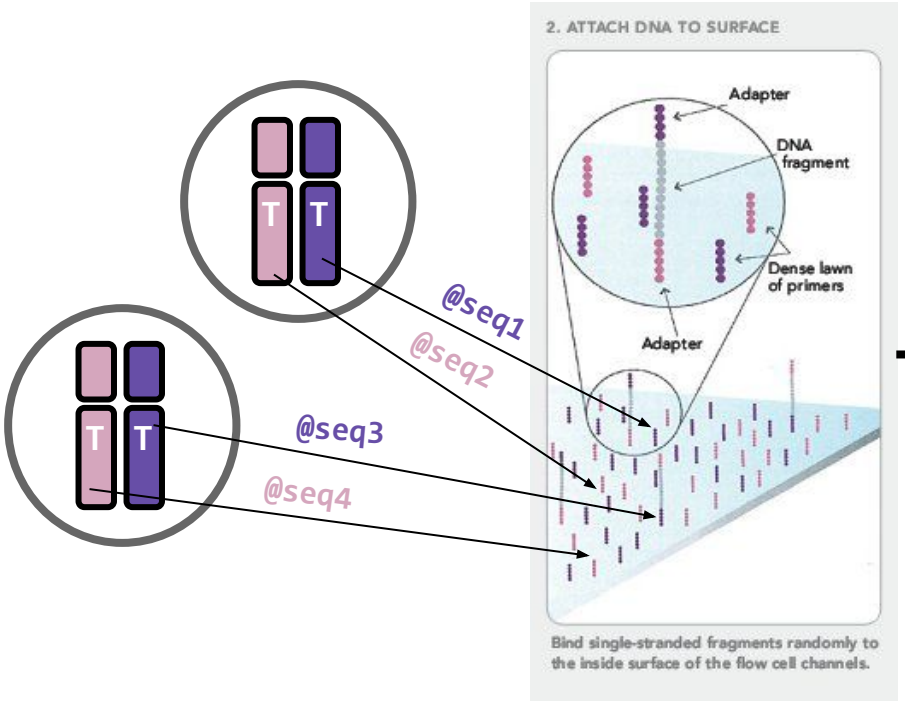


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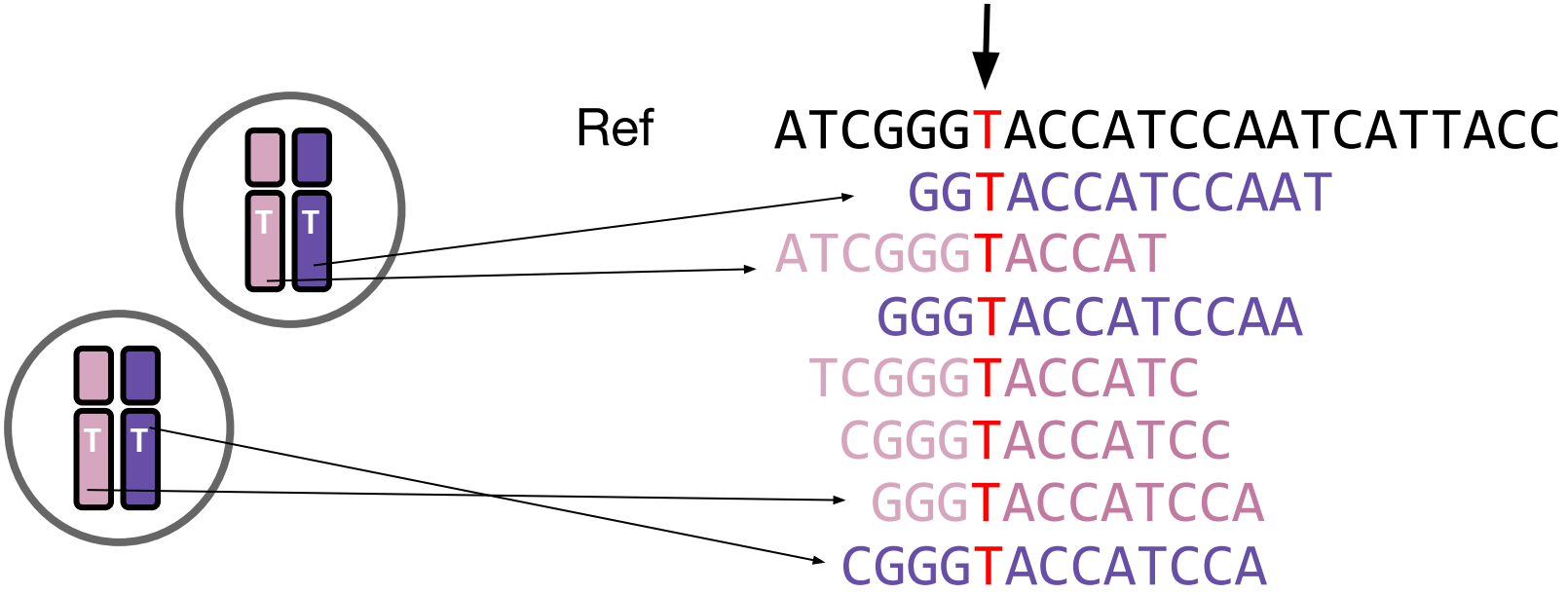
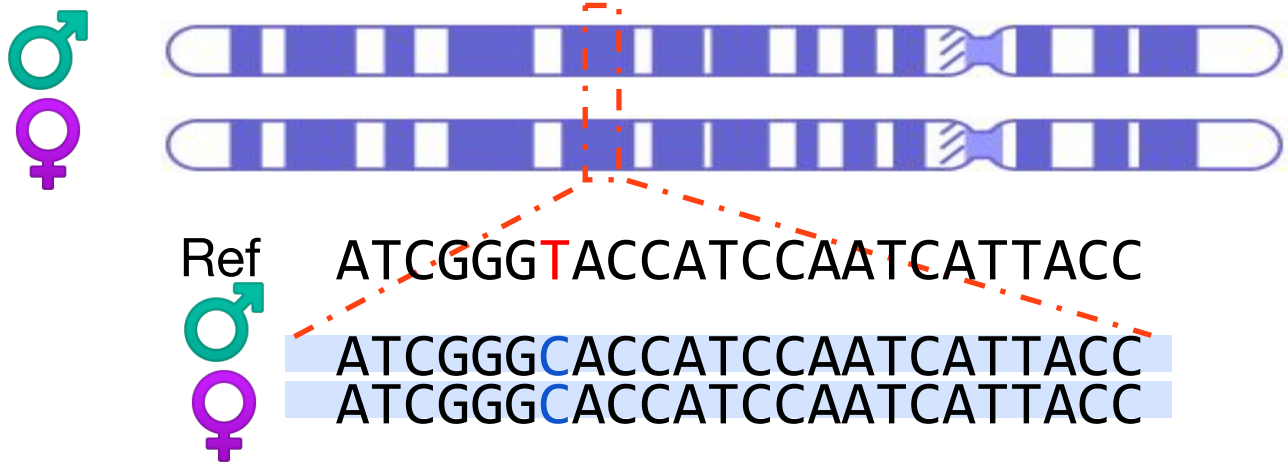


# Scenario 2: An individual is homozygous for an "alternate" allele.



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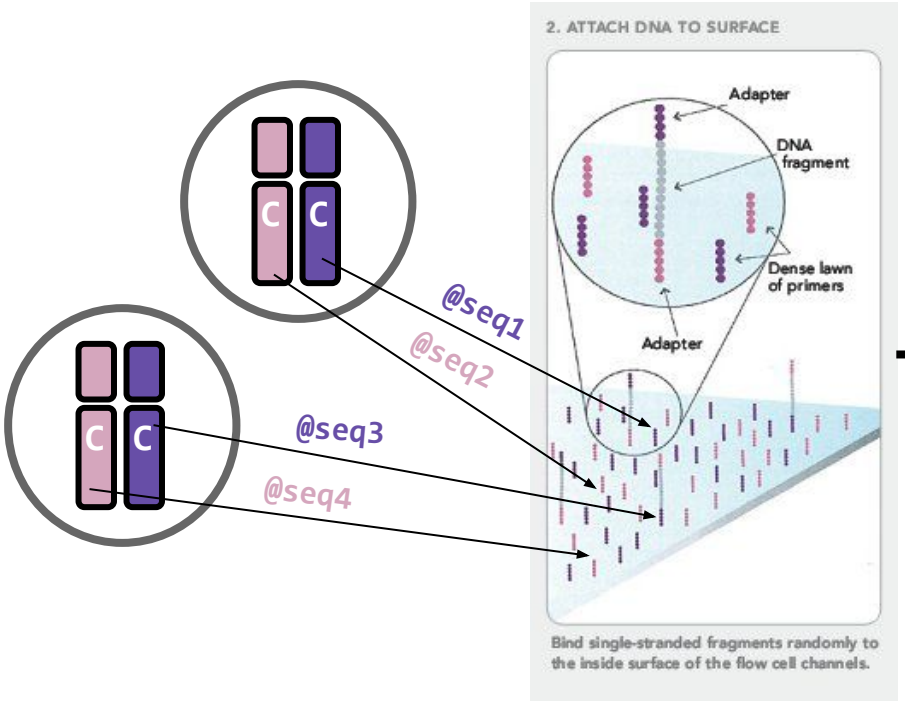





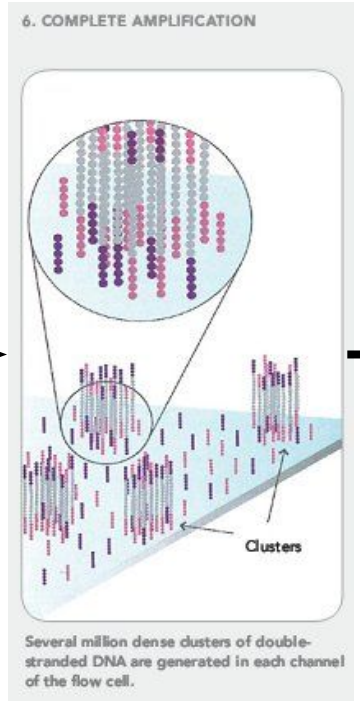


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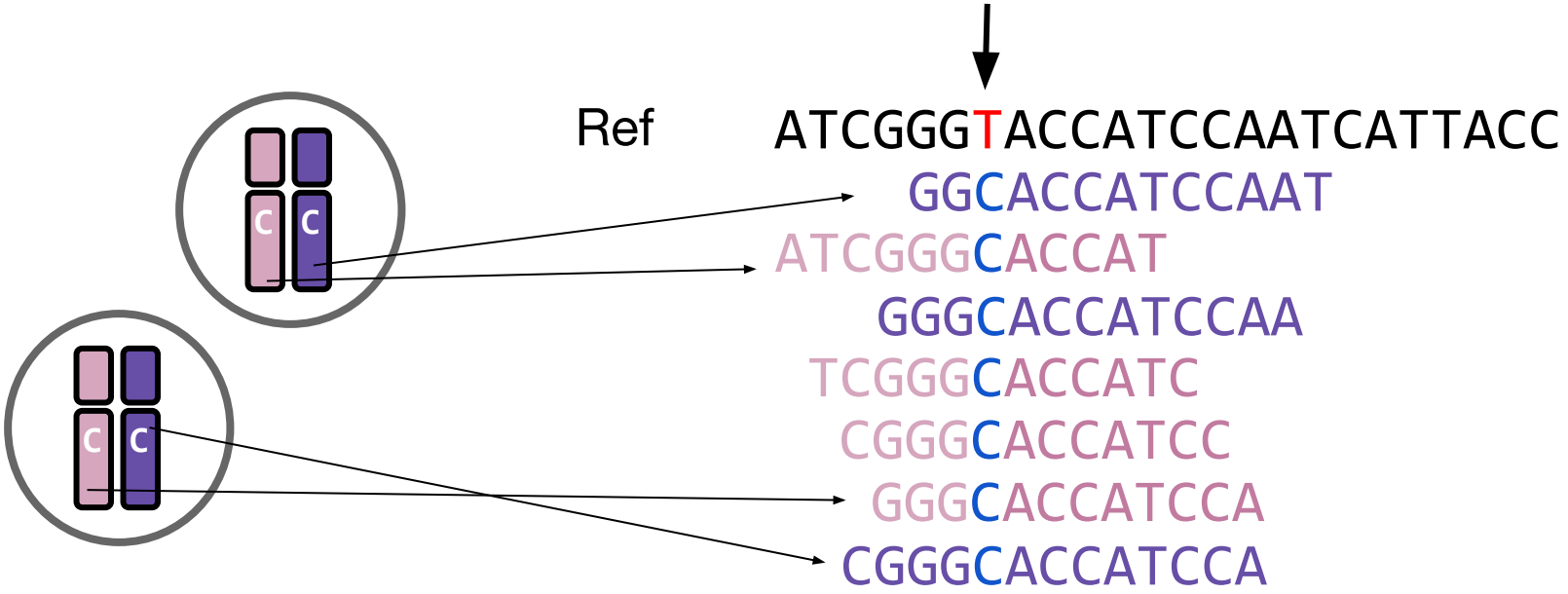
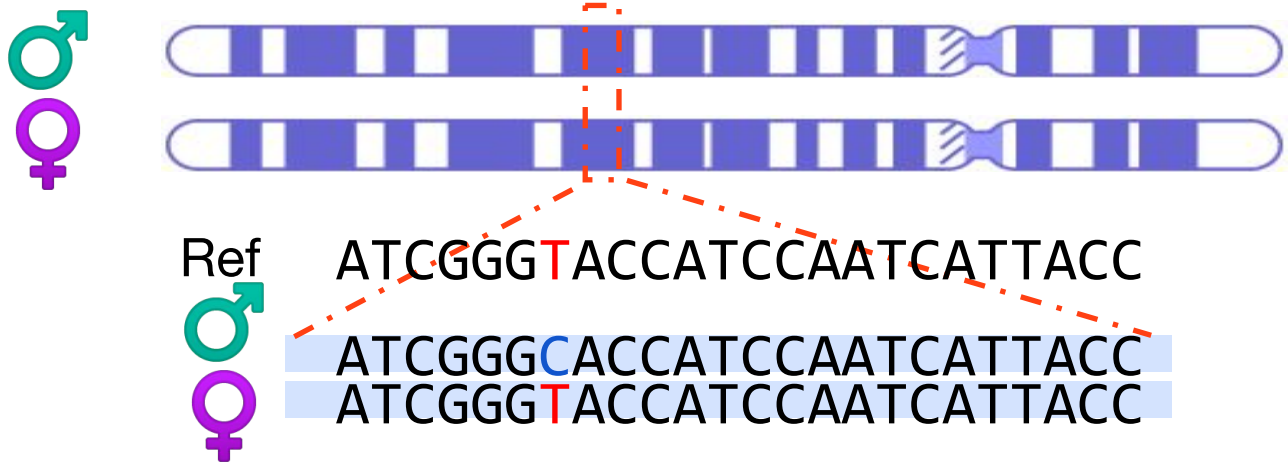


# Scenario 3: An individual is heterozygous for an "alternate" allele.



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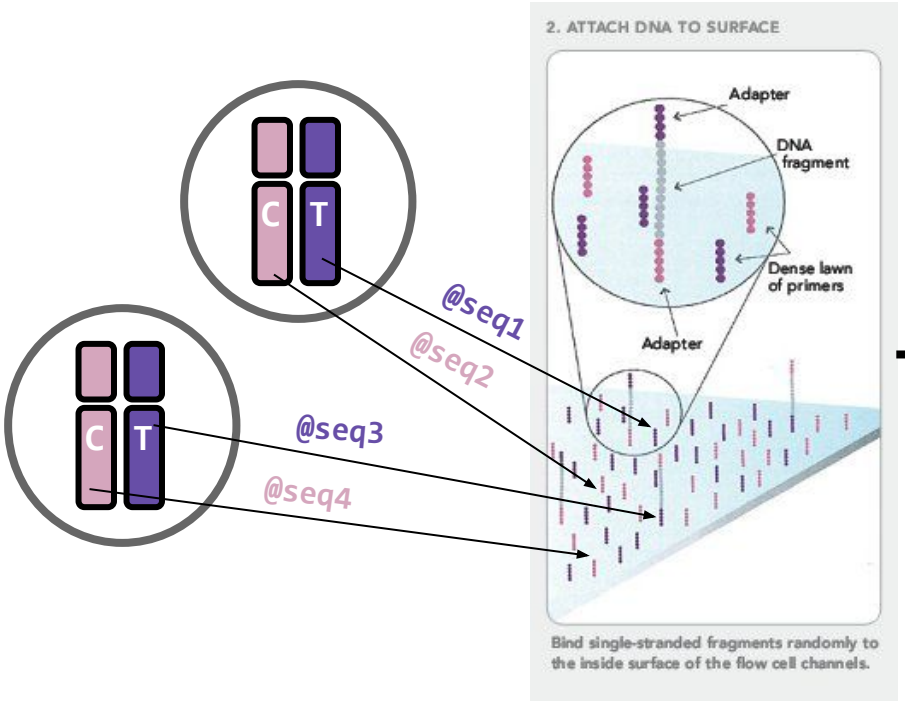





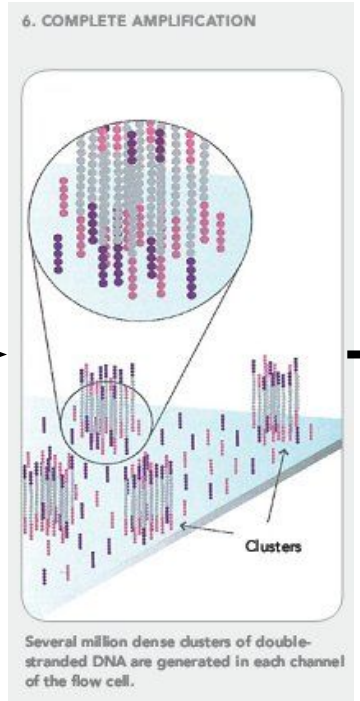


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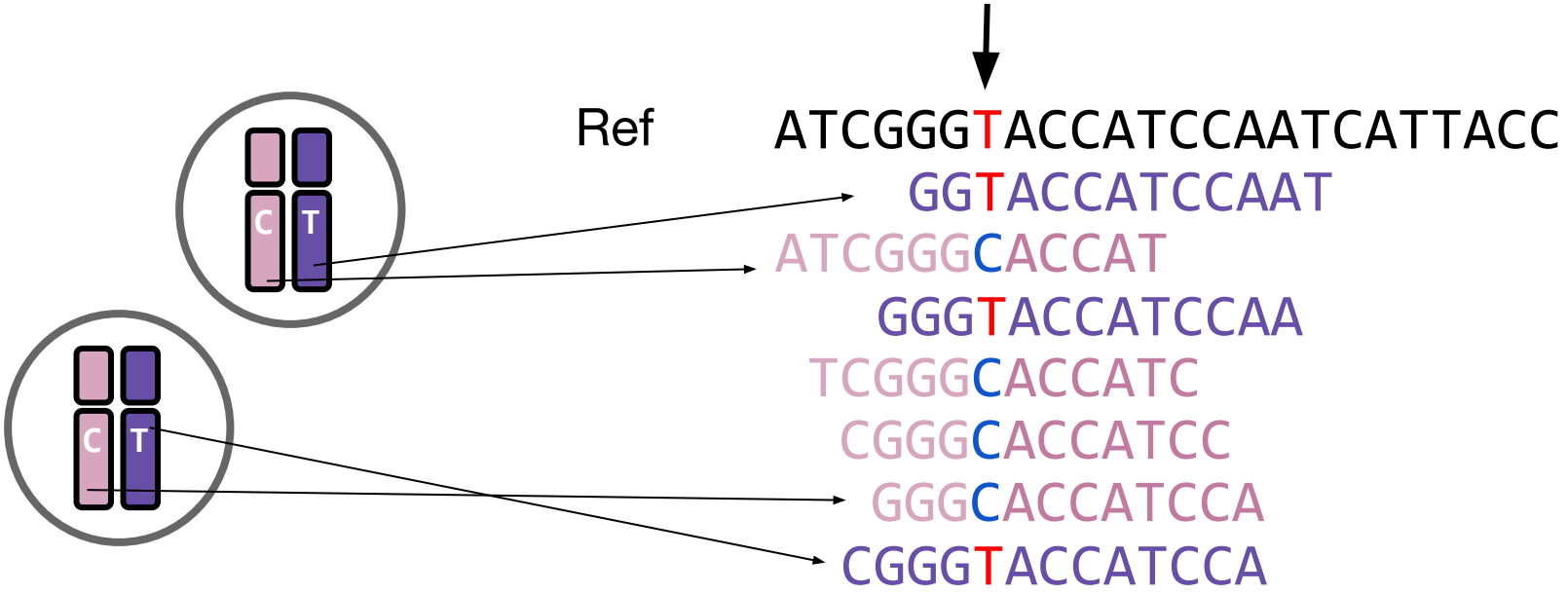

# Why might finding heterozygous variants be harder?

# The binomial distribution: adventures in coin flipping



P(heads) = 0.5

P(tails) = 0.5

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