Dizi Yerlestirme ve Varyant Tespiti Pratigi

Tugce Bilgin Sonay

- 156 kisinin mitokondriyal DNAsi
- Agizdan veya kandan ornek
- 20,000X dizi derinligi

PNAS

Maternal age effect and severe germ-line bottleneck in the inheritance of human mitochondrial DNA

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Footnotes

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Author contributions: B.R.-J., M.S.-W.S., F.C., R.N., M.M.H., I.M.P., A.N., and K.D.M. designed research; B.R.-J., M.S.-W.S., N.S., J.A.M., and B.D. performed research; D.B., T.S.K., and R.N. contributed new reagents/analytic tools; I.M.P. organized sample collection; B.R.-J., M.S.-W.S., N.S., and K.D.M. analyzed data; and B.R.-J., M.S.-W.S., N.S., F.C., M.M.H., A.N., and K.D.M. wrote the paper.

The authors declare no conflict of interest.

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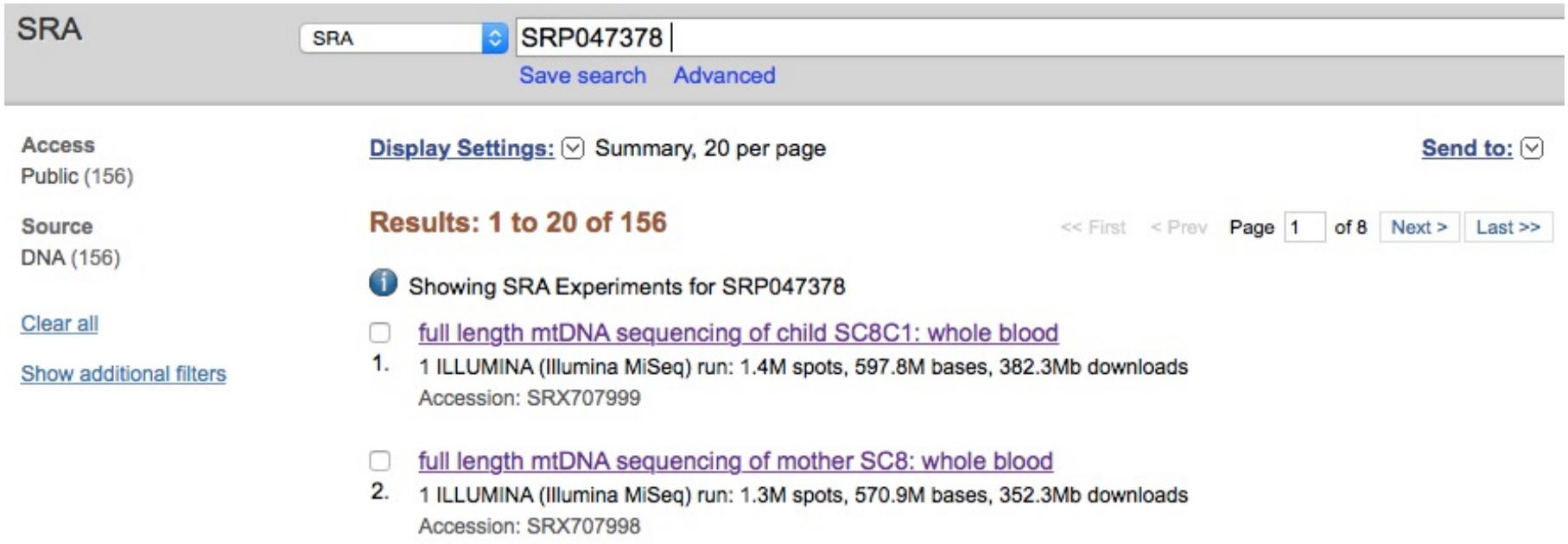
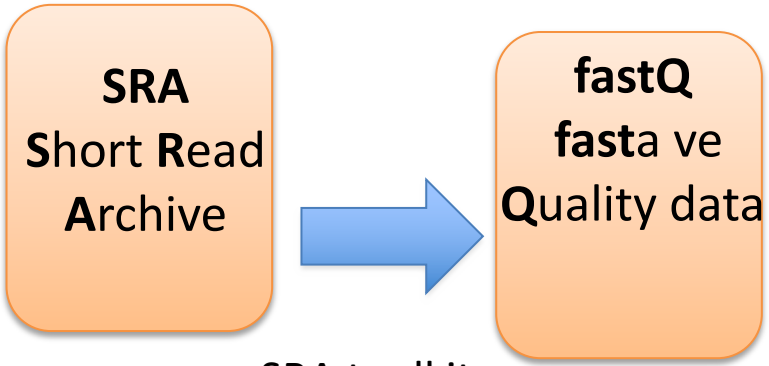
Data deposition: The sequences reported in this paper have been deposited in the Sequence Read Archive, www.ncbi.nlm.nih.gov/sra (accession no. SRP047378).

This article contains supporting information online at www.pnas.org/lookup/suppl/doi:10.1073/pnas.1409328111/-/DCSupplemental.

SRA
Short **R**ead
Archive





- Gene ID, uzunluk
- Fasta dizisi
- Gene ID, uzunluk
- Kalite verisi

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GGTTCCTACTTCAGGGTCATAAAGCCTAAATAGCCCACACGTTC
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<????BBBDDDDDDDDDGGGGGGIHIIIIIIIIHIIIIIHHHHII
```

Dizinin kalite degerleri

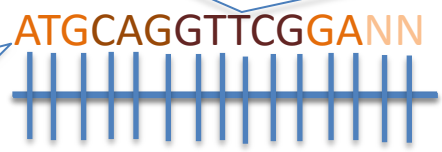
kalite degeri: 30 -> baz %99.9 ihtimal dogru

ATGCAGGTTCGGANN

Dizinin kalite degerleri

kalite degeri: 30 -> baz %99.9 ihtimal dogru

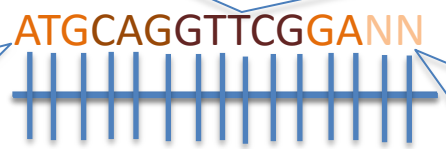
kalite degeri: 10-> baz %10 yanlis.



Dizinin kalite degerleri

kalite degeri: 30 -> baz %99.9 ihtimal dogru

kalite degeri: 10-> baz %10 yanlis.



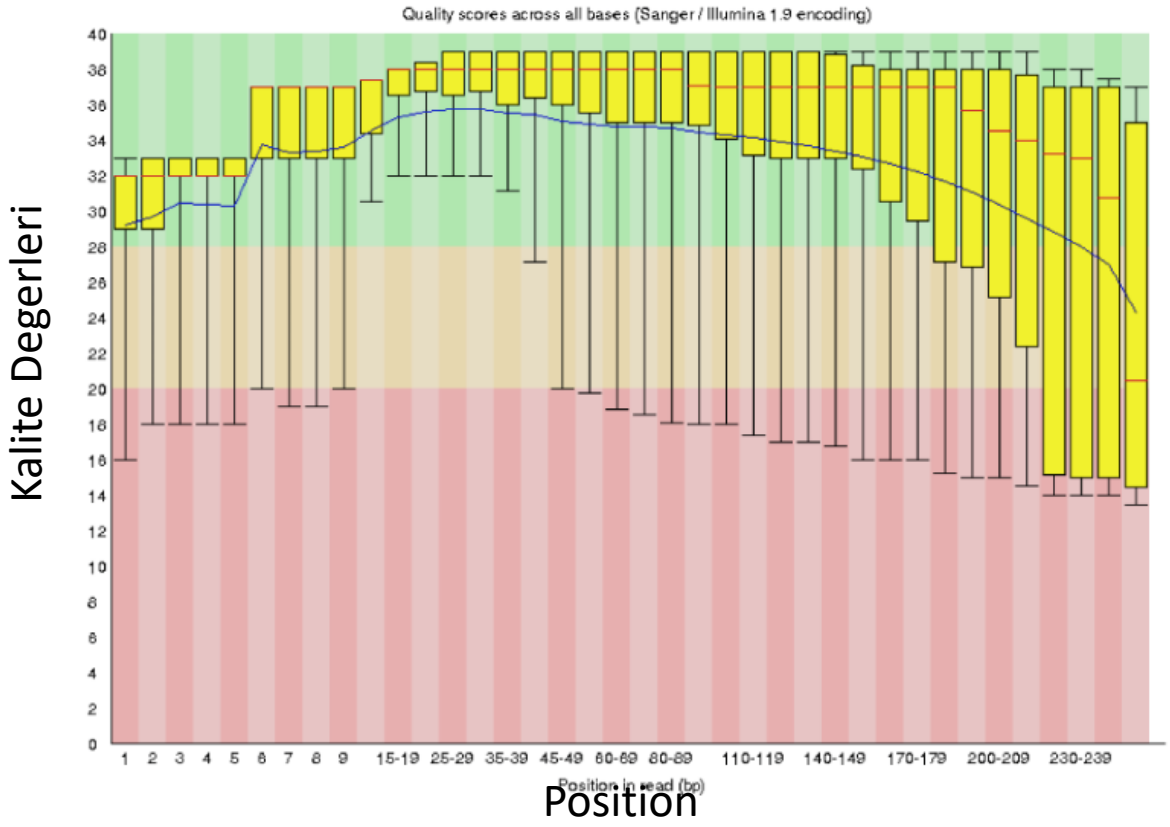
bazin ne oldugu belirlenemediyse N

SRA
Short **R**ead
Archive

SRA toolkit

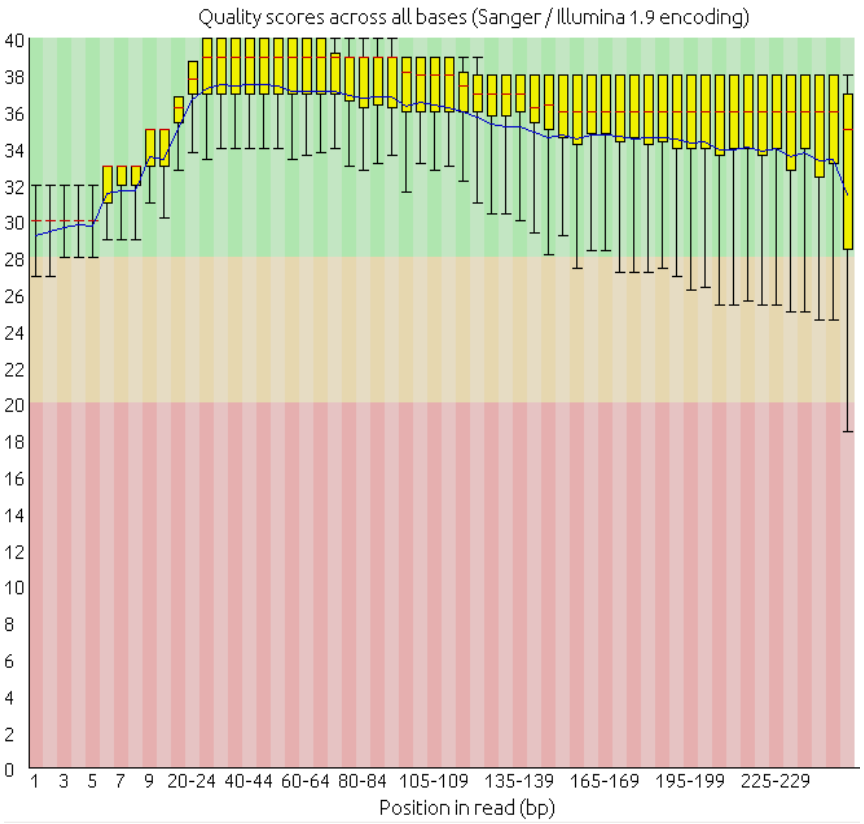
fastQ
fasta ve
Quality data

FastQC

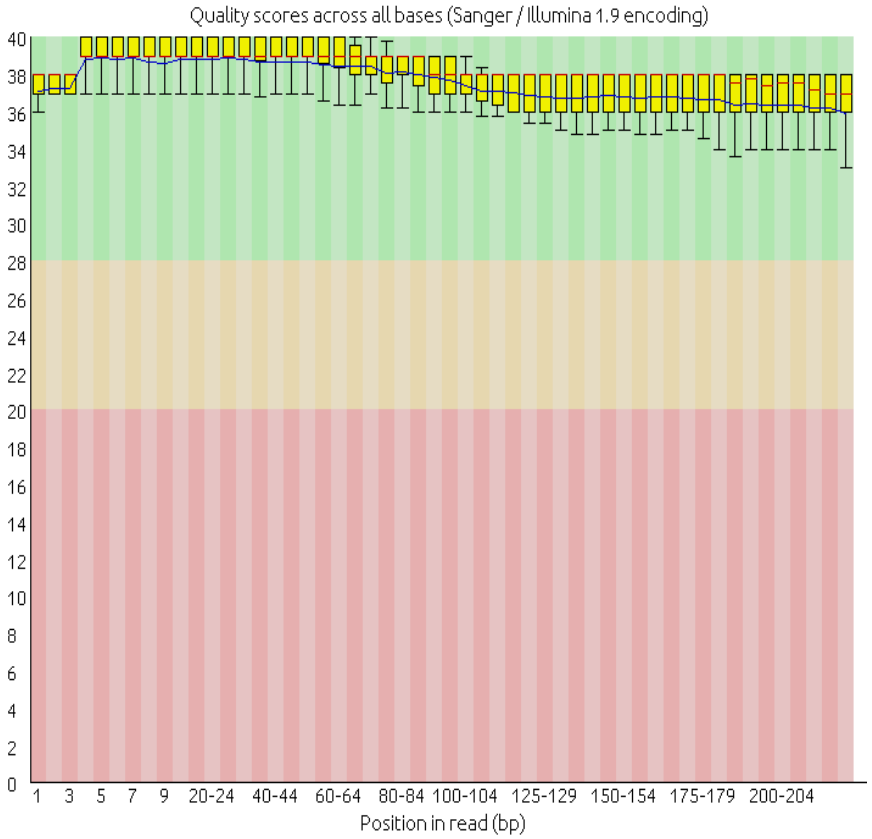


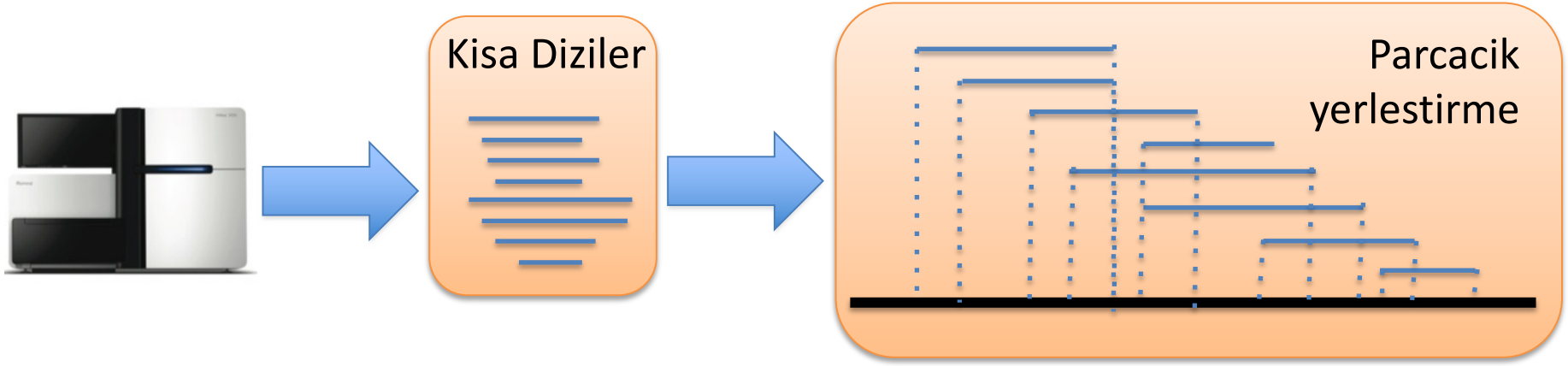
- Bastaki ve sondaki kotu kalite bazlari kes
- Aralardaki kotu kalite bazlari filtrele

Oncesi

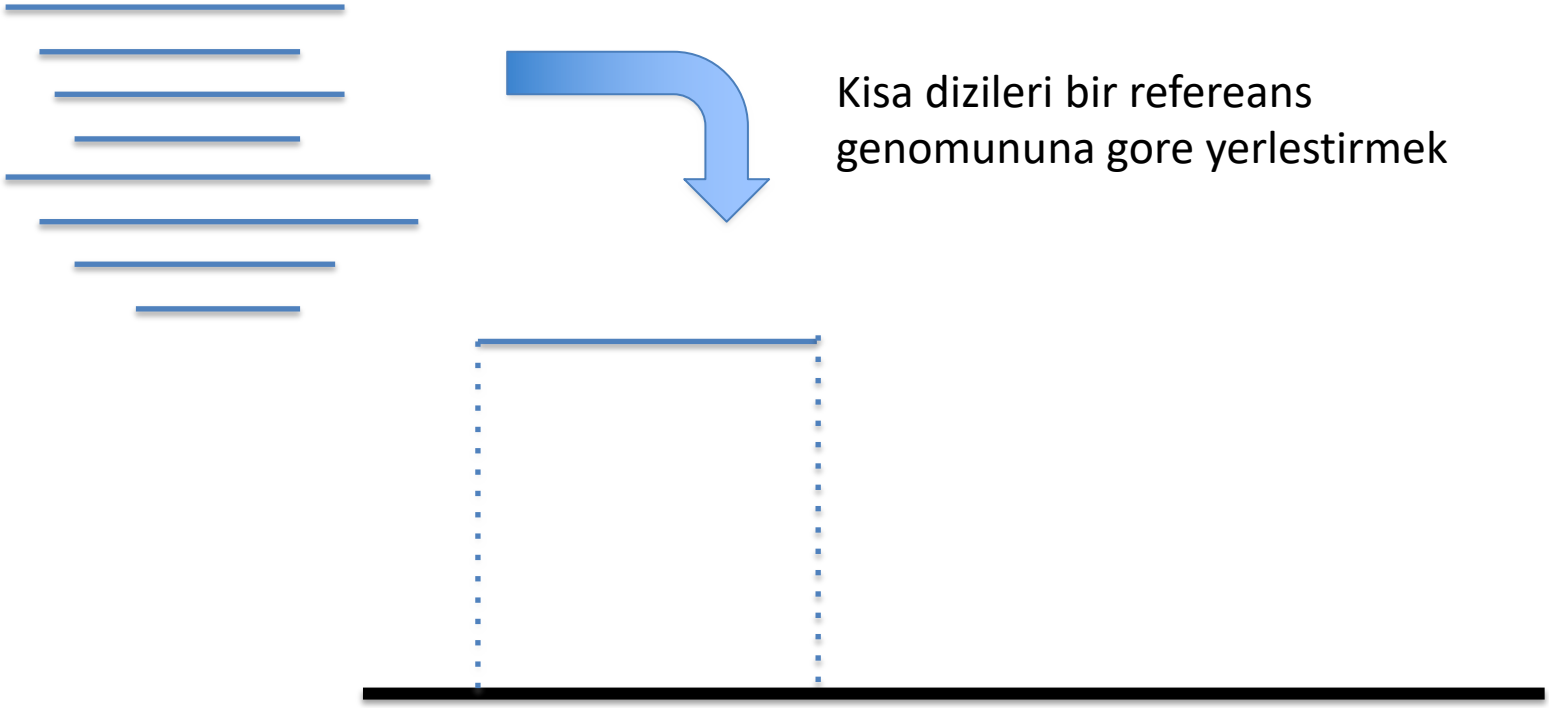


Sonrasi



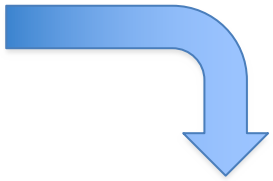

Kisa Diziler
Parcacik
yerlestirme

Parcacik Yerlestirme



Kisa dizileri bir refereans genomununa gore yerlestirmek

Parcacik Yerlestirme



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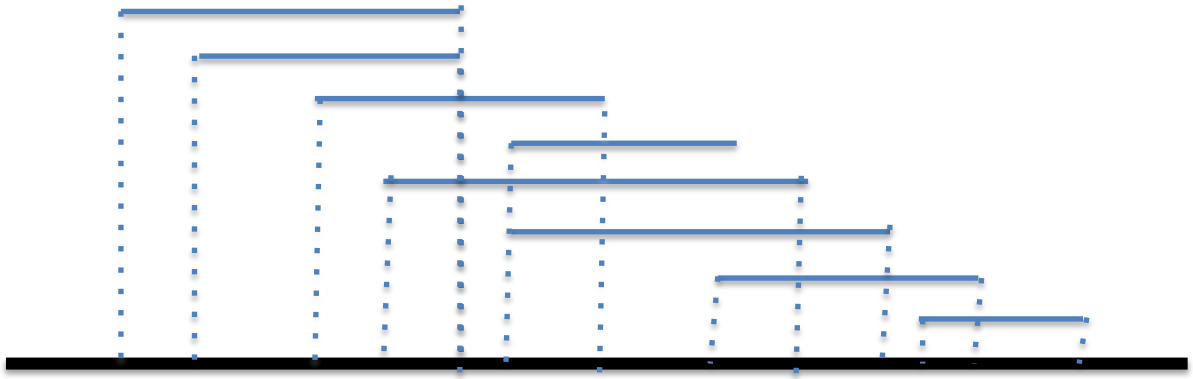
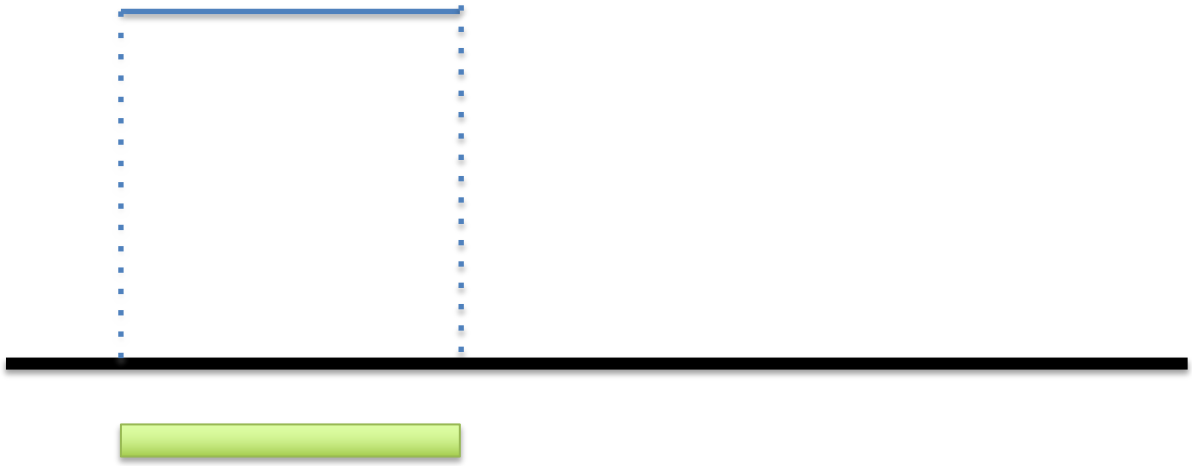


Yerlestime Kalitesi



Yerlestime Kalitesi

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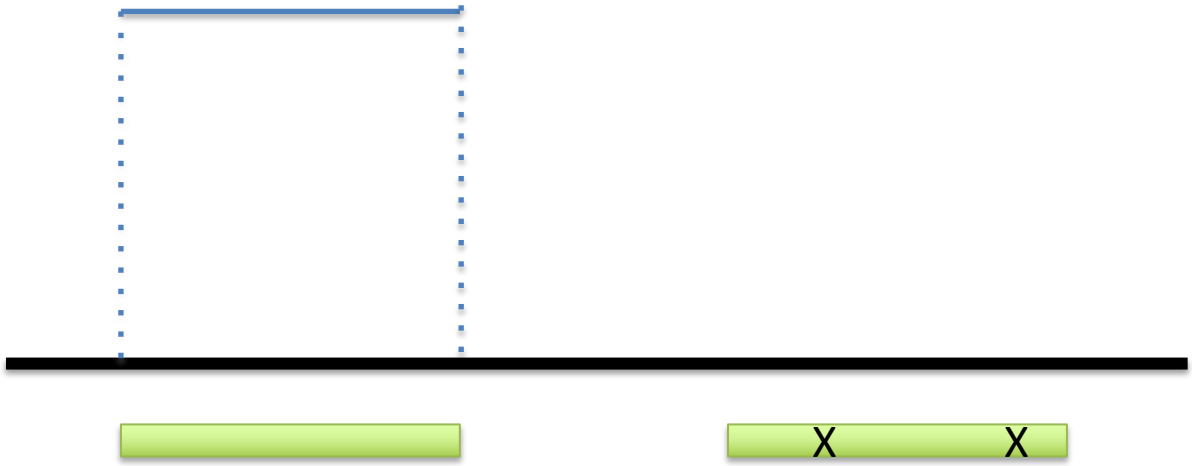
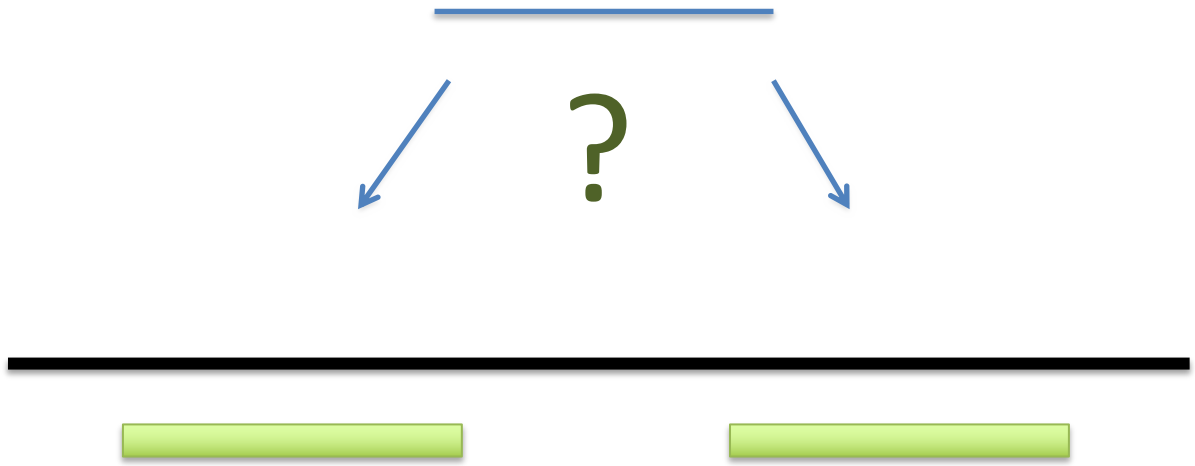
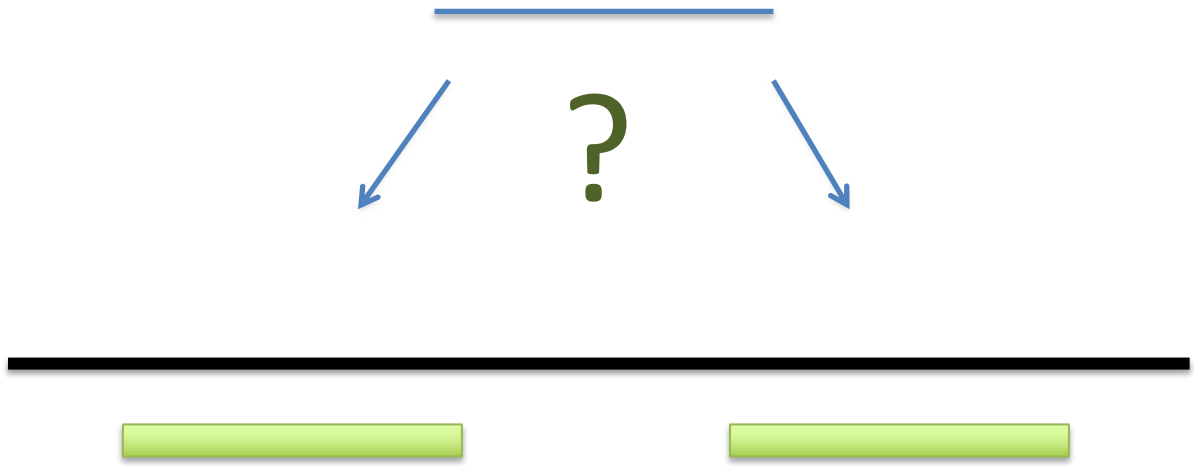


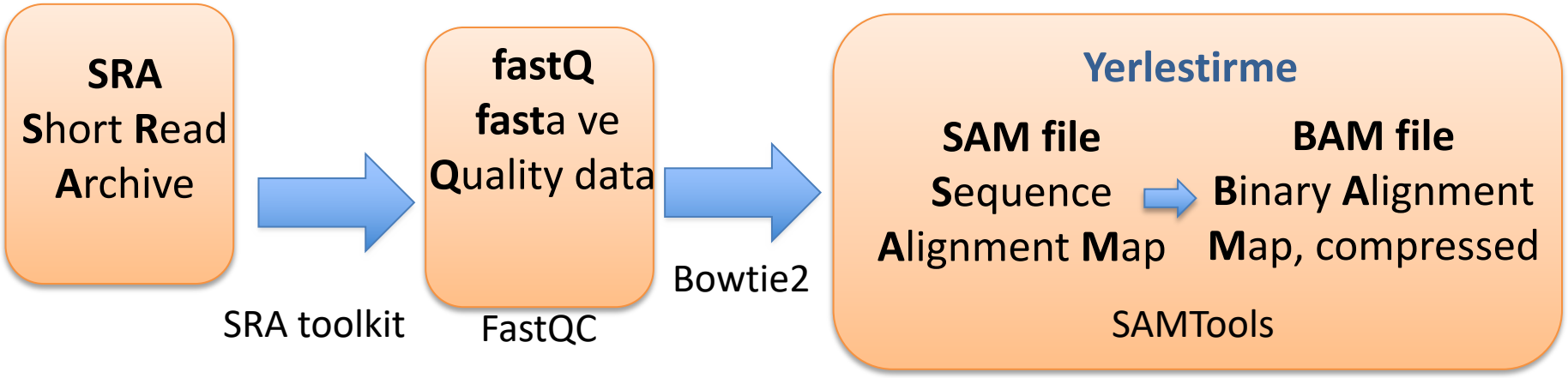
Yerlestime Kalitesi



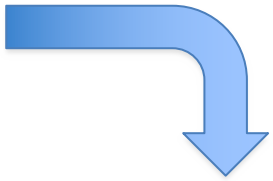
Yerlestime Kalitesi

Bu parcacik genoma yerlestirilirken ne kadar kesin konusabiliriz?

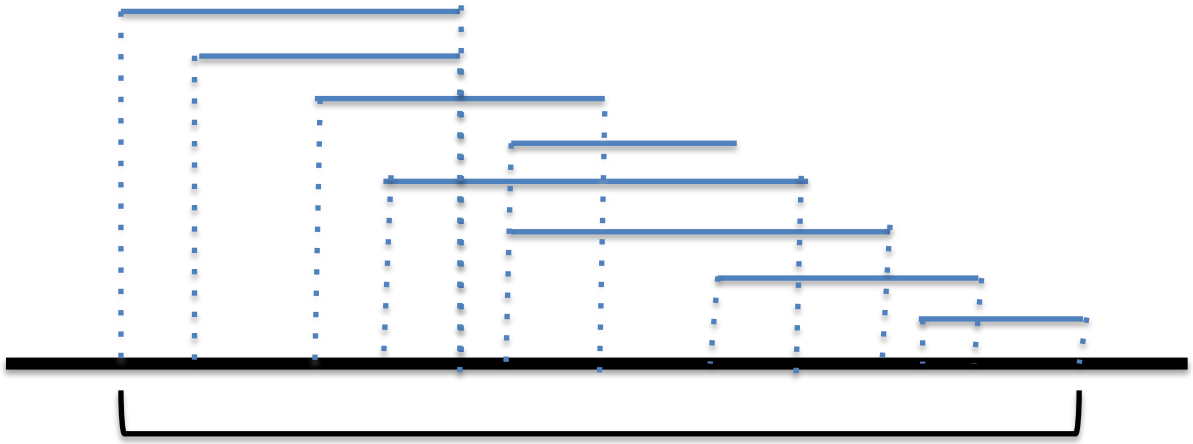



SRA
Short Read
Archive
SRA toolkit
fastQ
fasta ve
Quality data
FastQC
Bowtie2
Yerlestirme
SAM file
Sequence
Alignment Map
BAM file
Binary Alignment
Map, compressed
SAMTools

Parcacik Yerlestirme

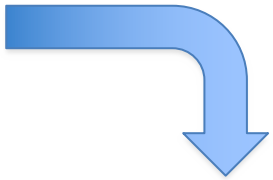


Kisa dizileri bir refereans genomununa gore yerlestirmek



Kapsanan Genom: %90

Parcacik Yerlestirme



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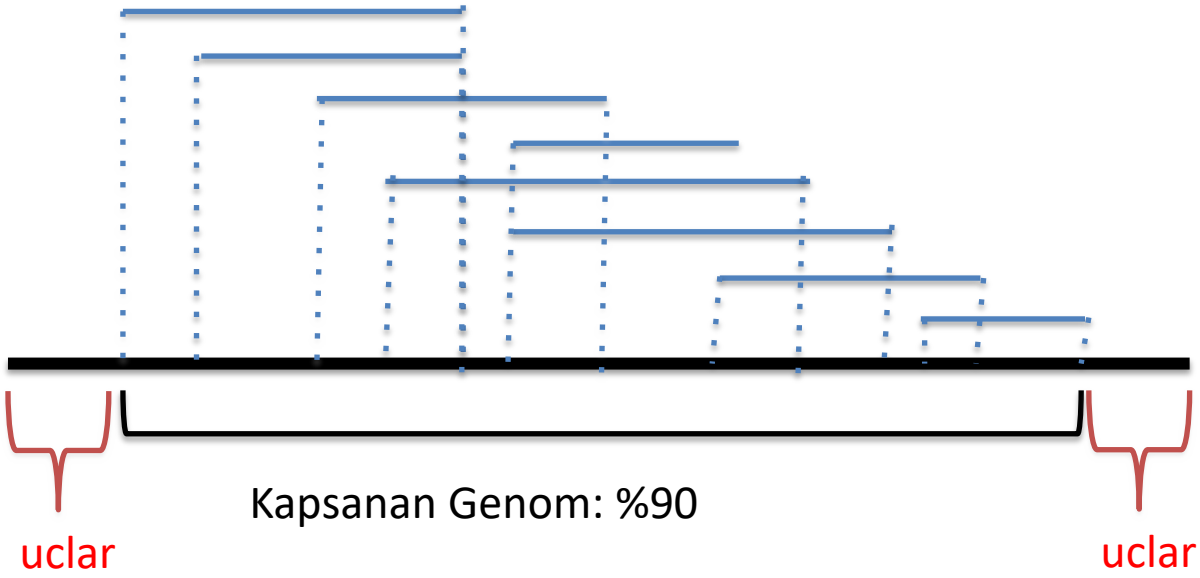
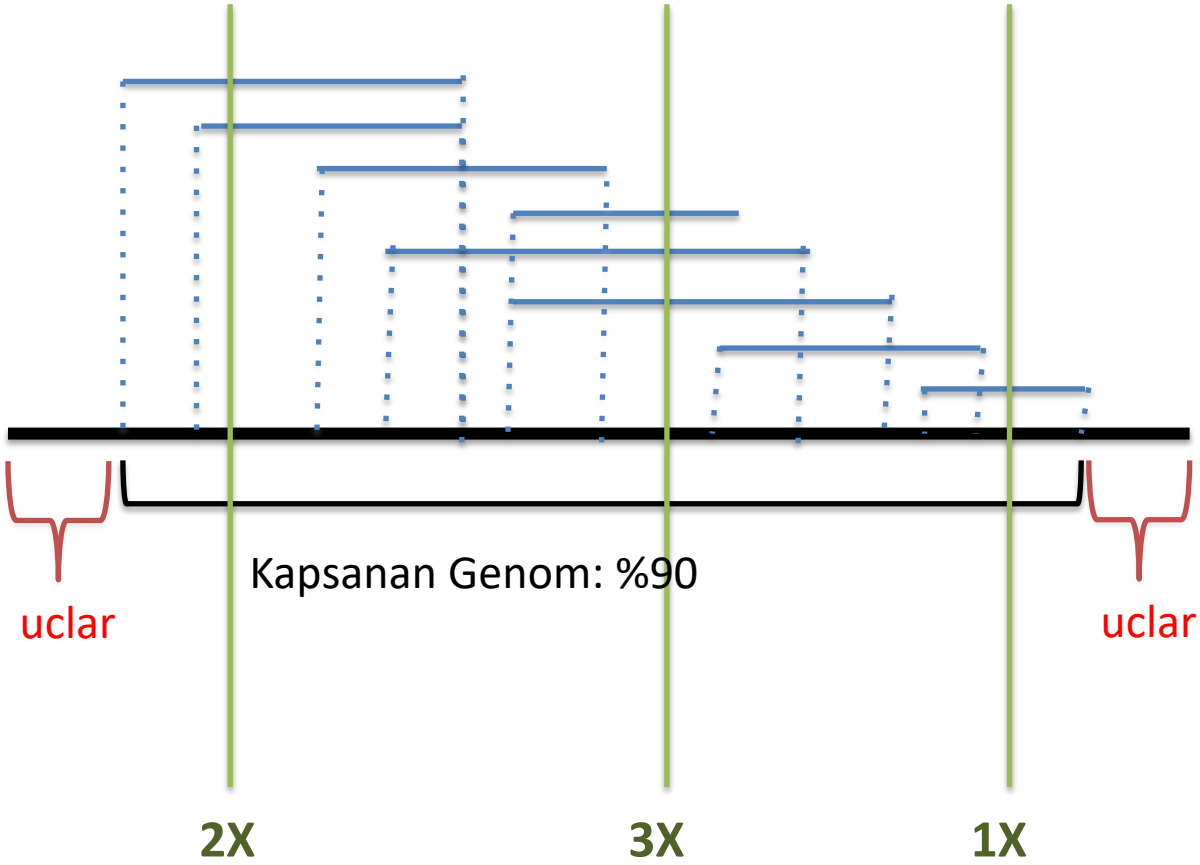
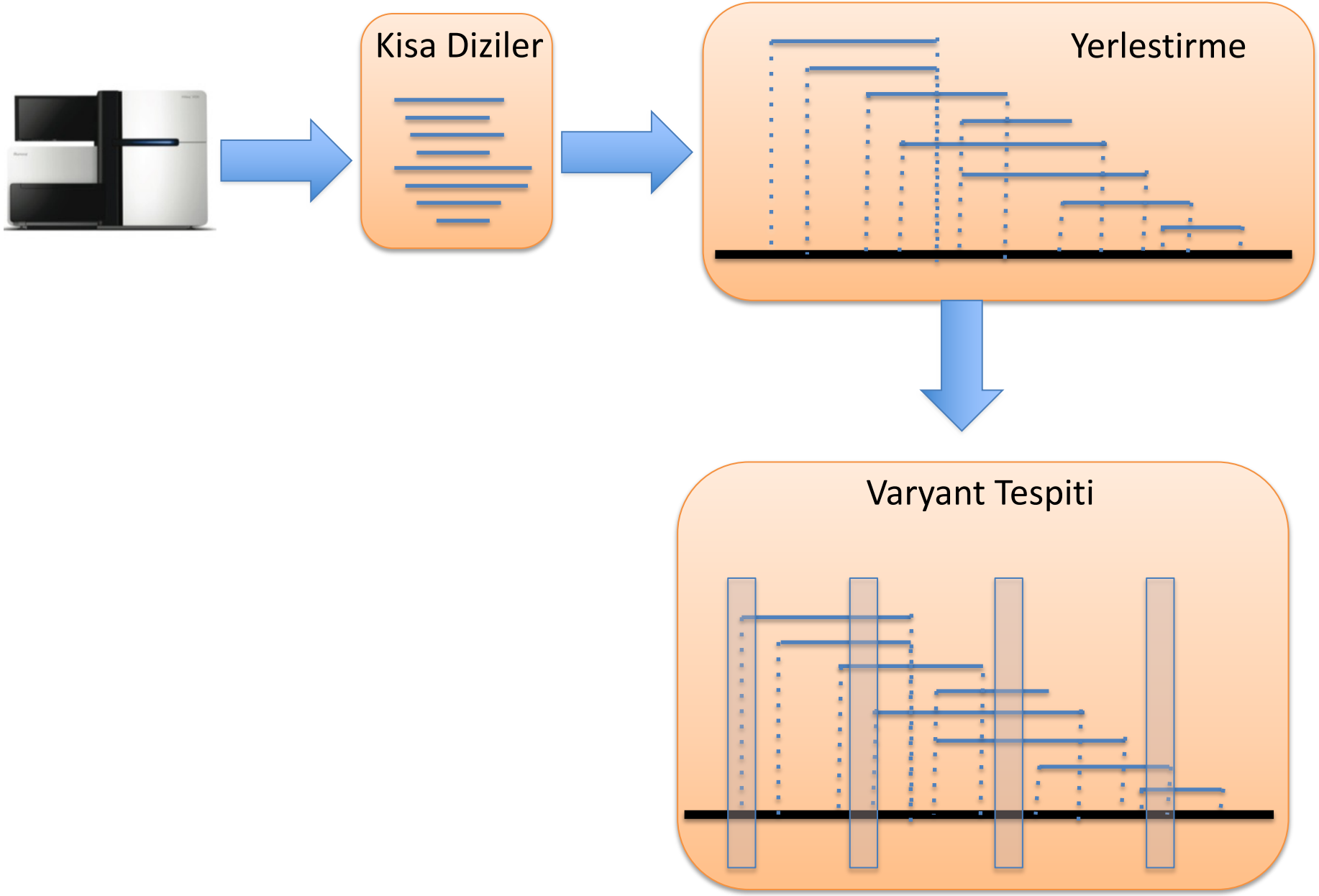


Parcacik Yerlestirme

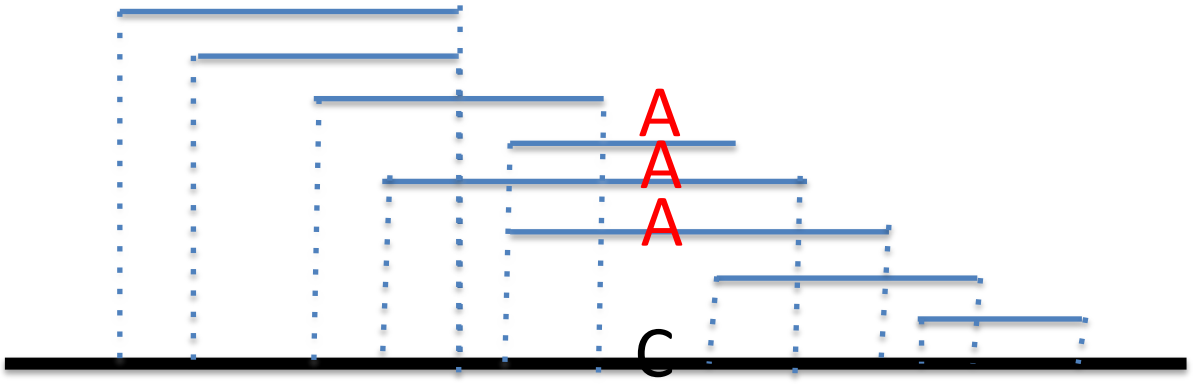
Kisa dizileri bir refereans genomununa gore yerlestirmek

Dizi Derinligi– bir baz kac kere yerlestiildi

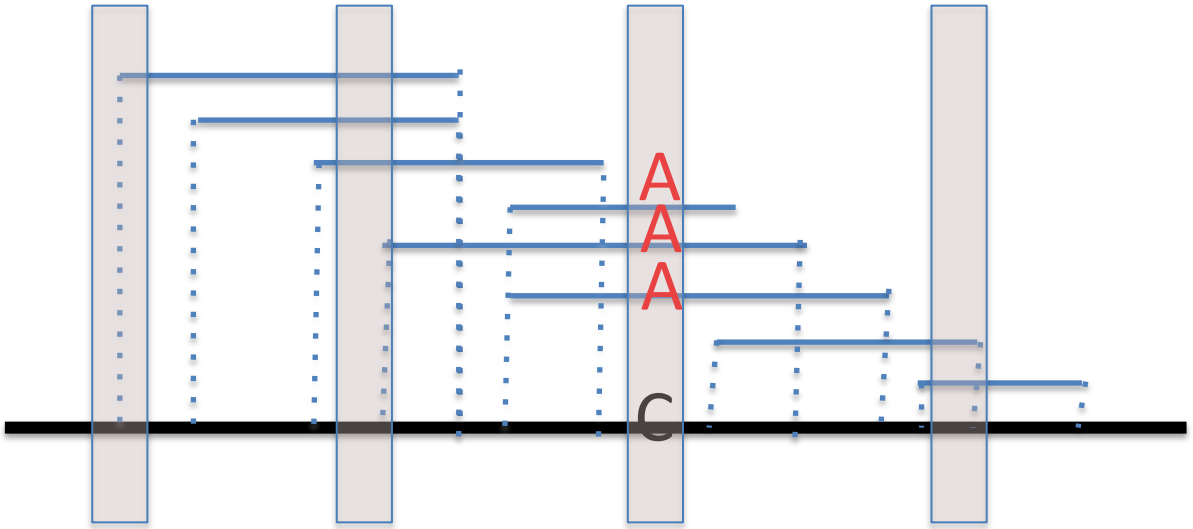


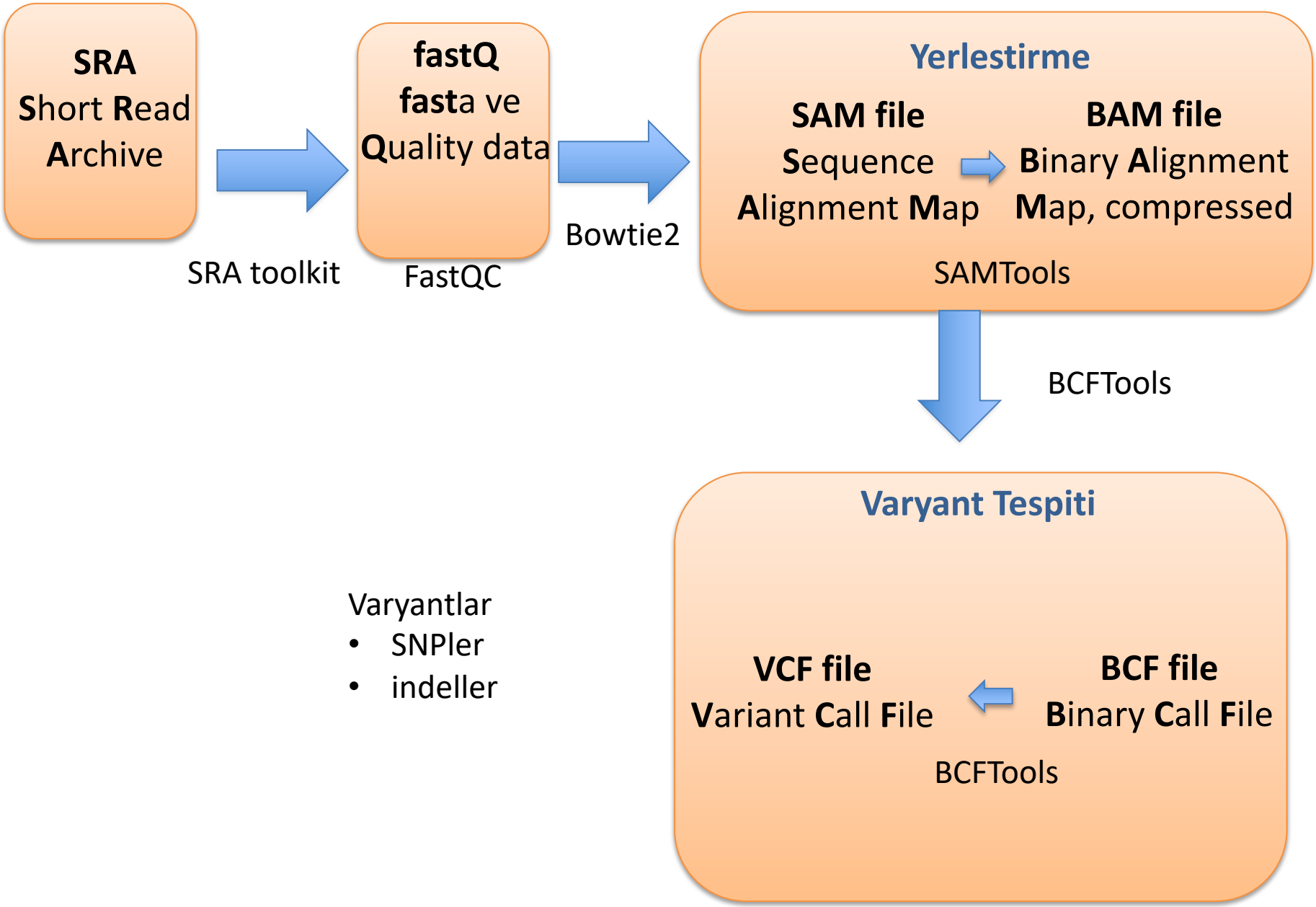

Kisa Diziler
Yerlestirme
Varyant Tespiti

Varyant Tespiti



Varyant Tespiti





Varyantlar

- SNPler
- indeller

Variant Call File Format