

Dizi Yerleştirme ve Varyant Tespiti Pratiği

Tugce Bilgin Sonay



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



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

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- 20,000X dizi derinliği

Footnotes

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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 Read Archive

SRA

Access
Public (156)

Source
DNA (156)

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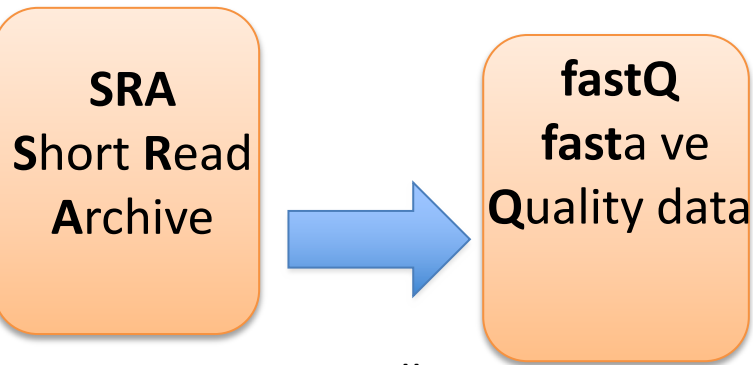
Send to: ☒

Results: 1 to 20 of 156

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 Showing SRA Experiments for SRP047378

- ☐ [full length mtDNA sequencing of child SC8C1: whole blood](#)
 1. 1 ILLUMINA (Illumina MiSeq) run: 1.4M spots, 597.8M bases, 382.3Mb downloads
Accession: SRX707999
- ☐ [full length mtDNA sequencing of mother SC8: whole blood](#)
 2. 1 ILLUMINA (Illumina MiSeq) run: 1.3M spots, 570.9M bases, 352.3Mb downloads
Accession: SRX707998



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

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?????BAADEEDEDDEDFGGGGGIIIIIIIIHIIIIIIHIIIIHIHI
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GGTTCCTACTTCAGGGTCATAAAGCCTAAATAGCCCACACGTTC
+SRR1583063.2.1 2 length=251
<?????BBBDDDDDDDDGGGGGGIIIIIIIIHIIIIIIHHHHII
```

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.

ATGCAGGTTCGGANN



Dizinin kalite degerleri

kalite degeri: 30 -> baz %99.9 ihtimal dogru

kalite degeri: 10-
> baz %10 yanlis.

ATGCAGGTTCGGANN



bazin ne oldugu
belirlenemediyse
N

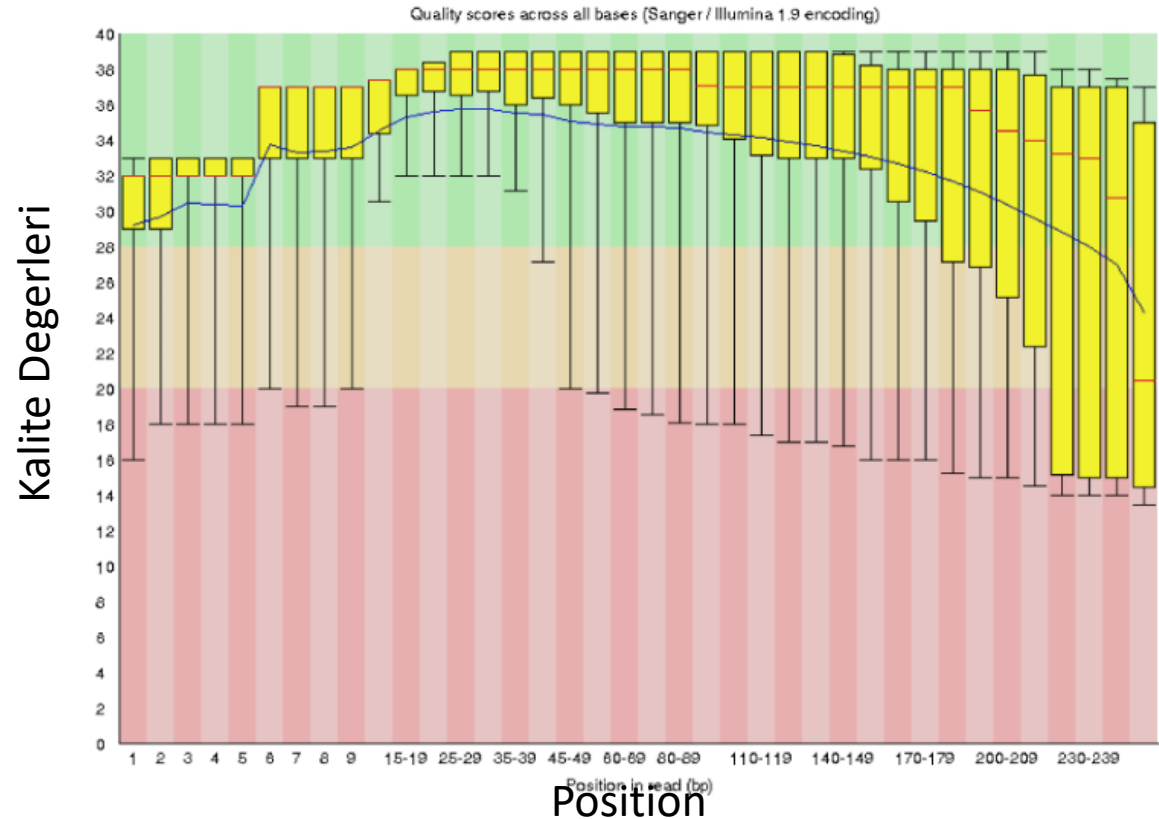
SRA
Short Read
Archive



fastQ
fasta ve
Quality data

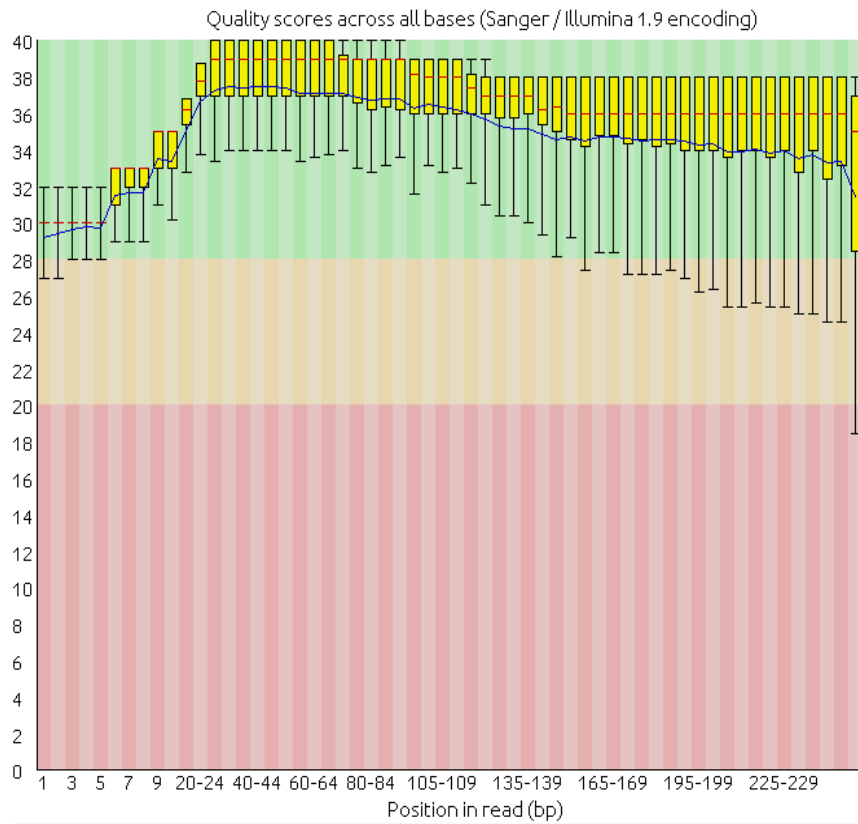
SRA toolkit

FastQC

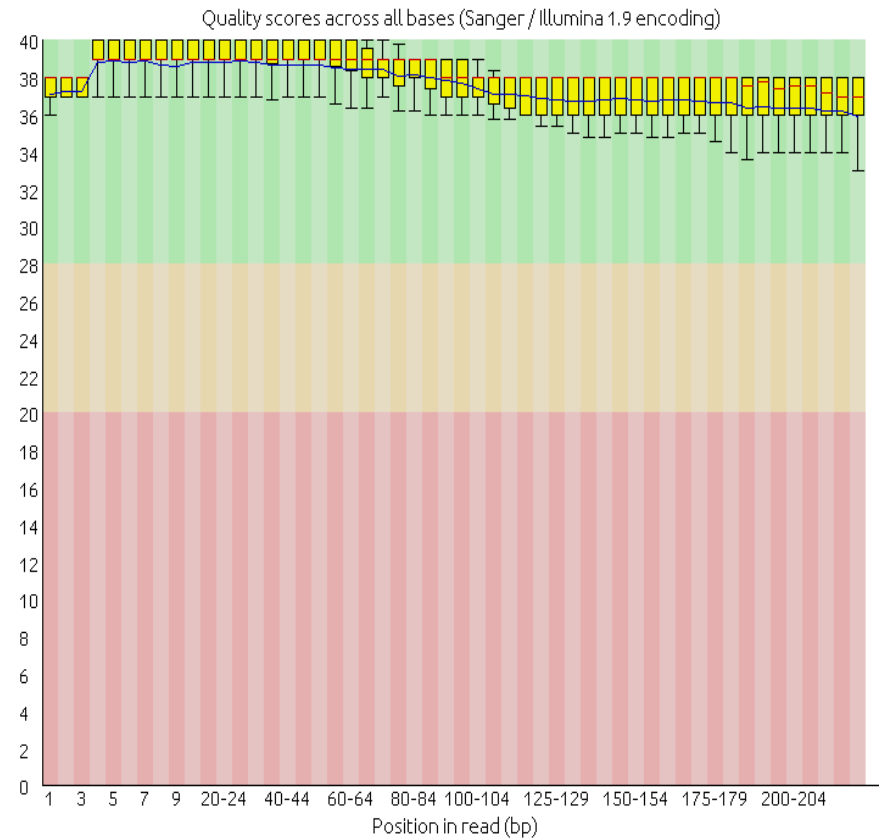


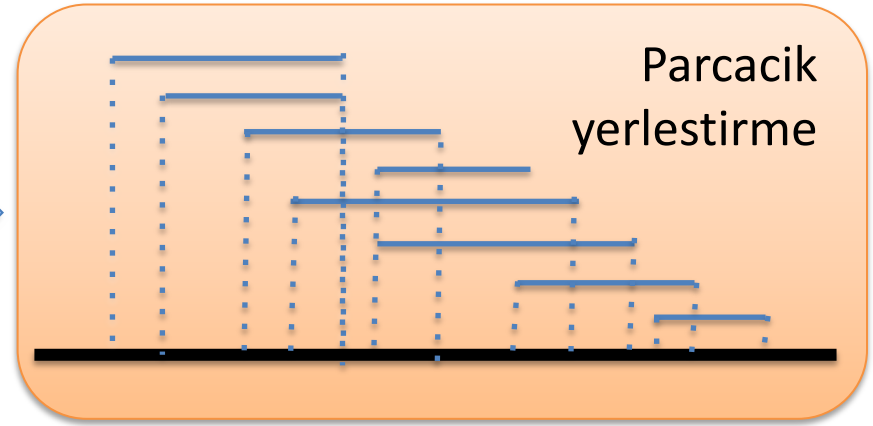
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- Aralardaki kotu kalite bazlari filtrele

Oncesi

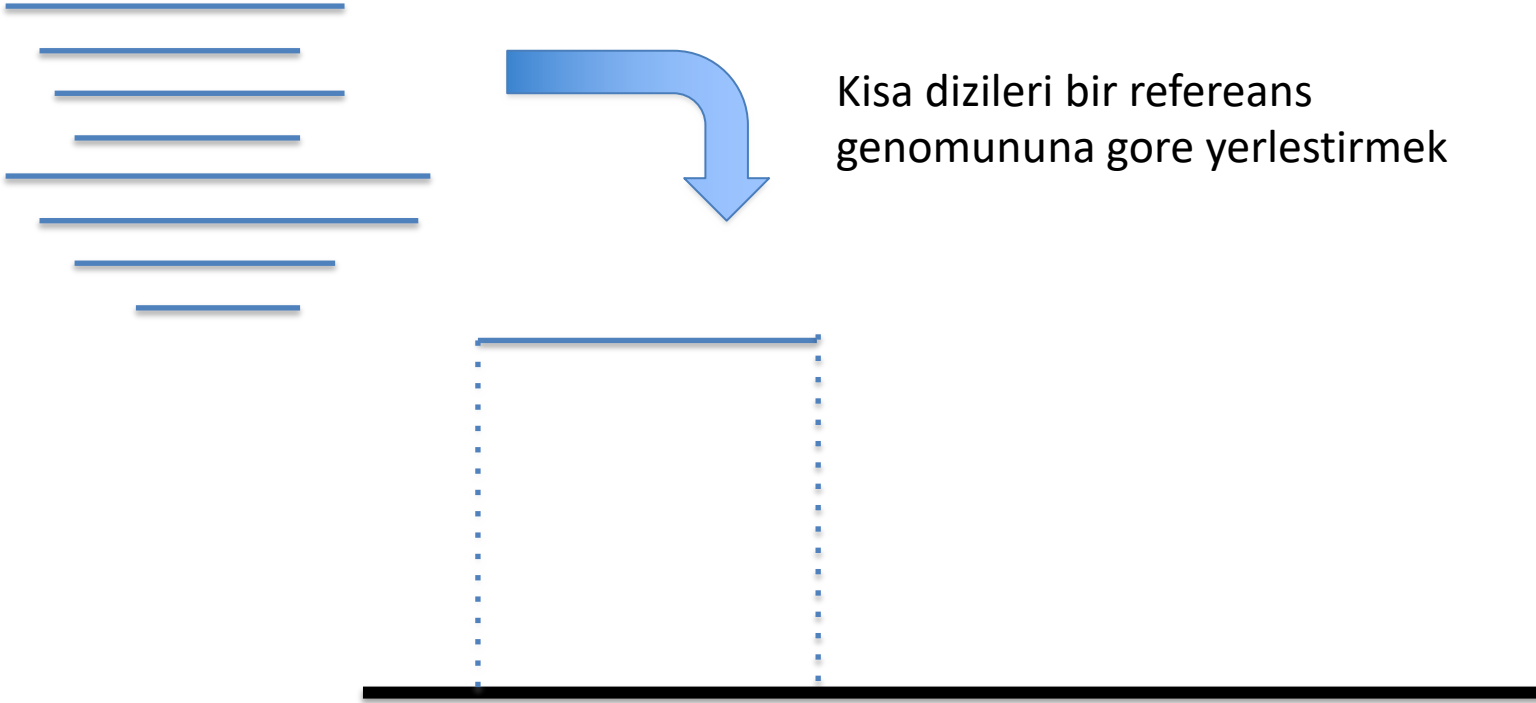


Sonrasi

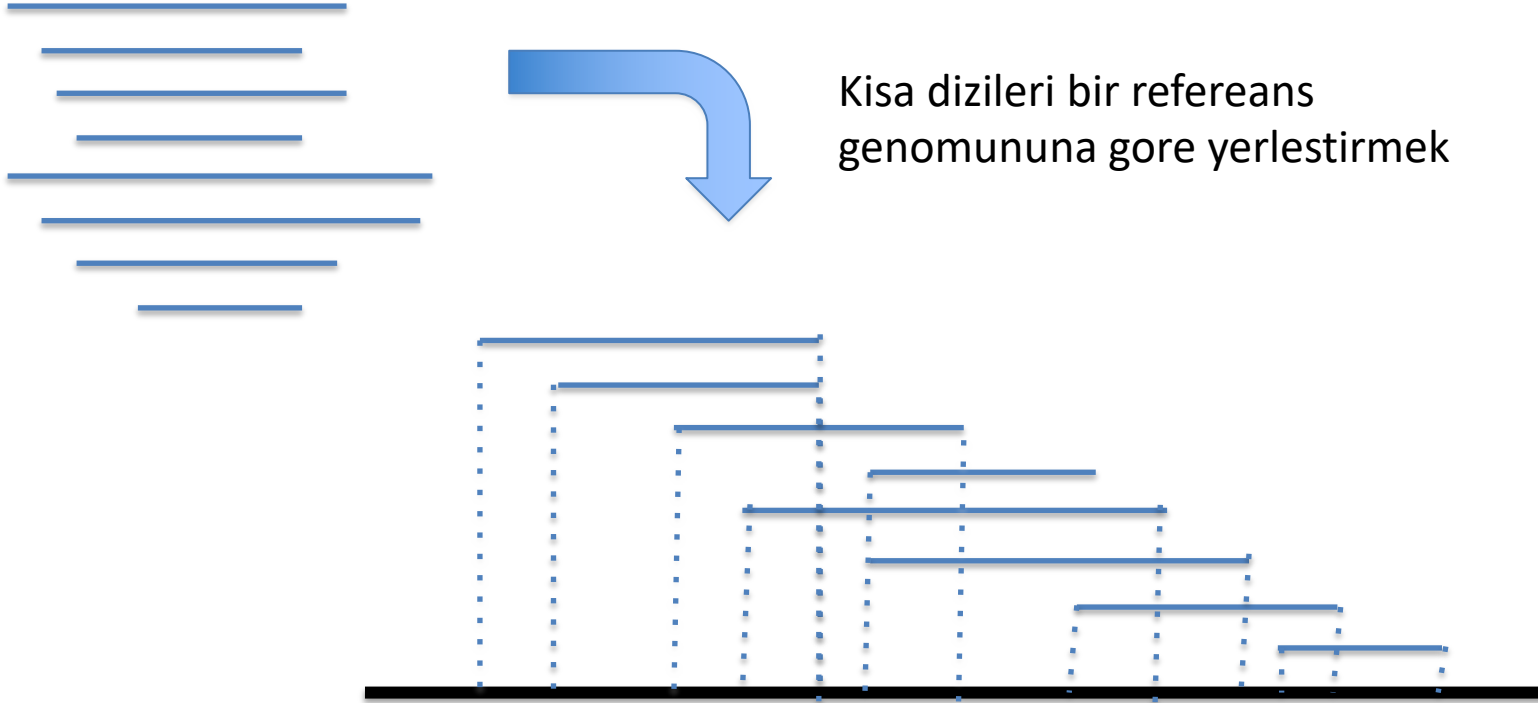




Parcacik Yerlestirme



Parcacik Yerlestirme



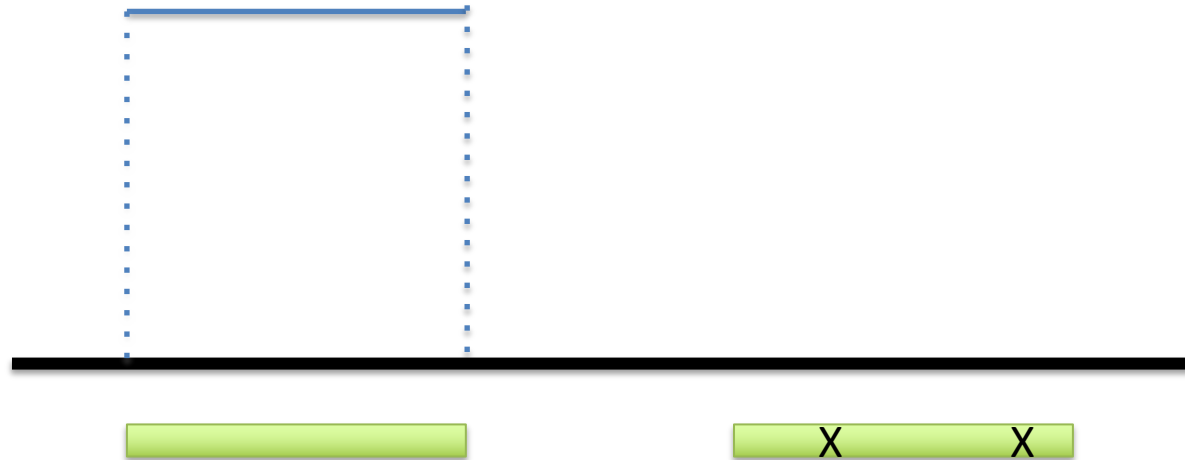
Yerlestim Kalitesi



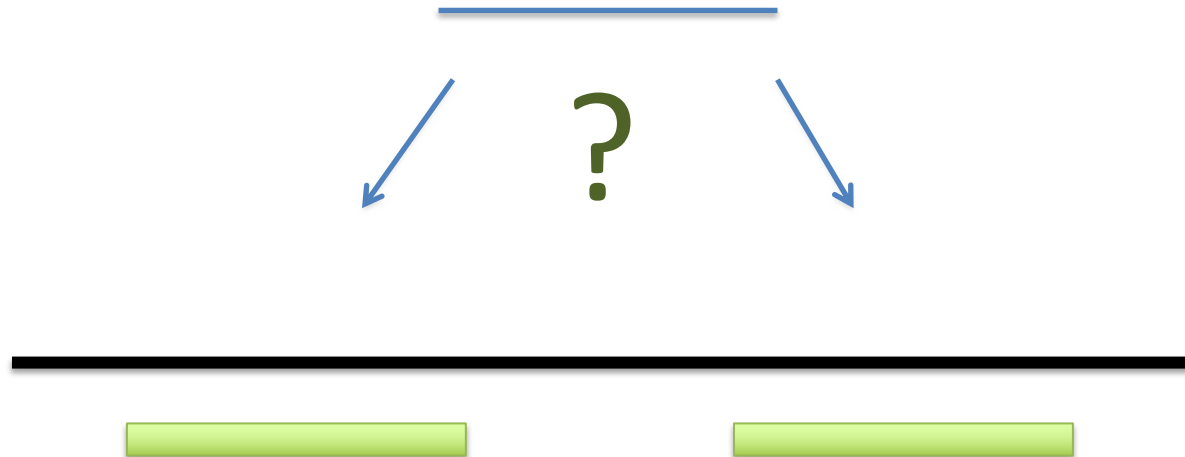
Yerleşttime Kalitesi



Yerleşttime Kalitesi

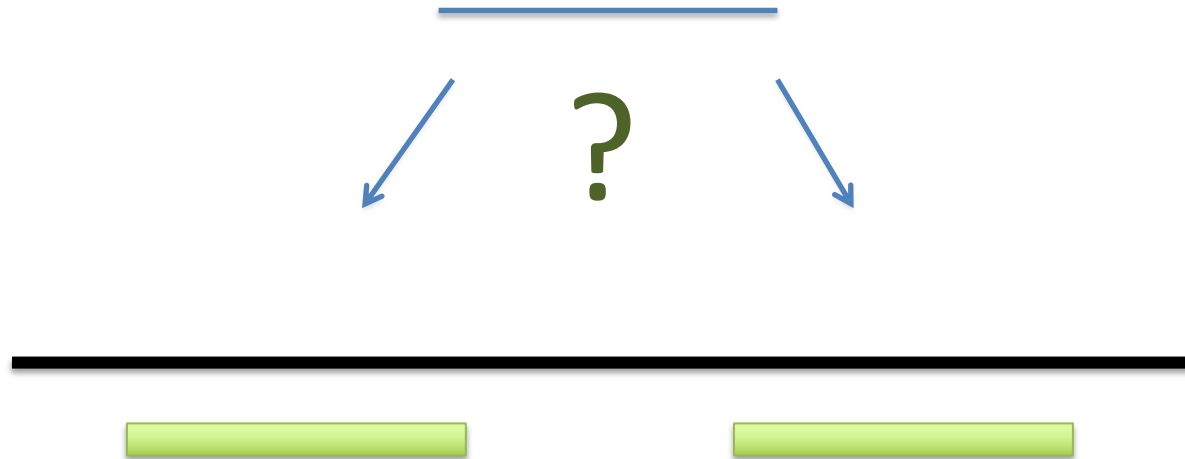


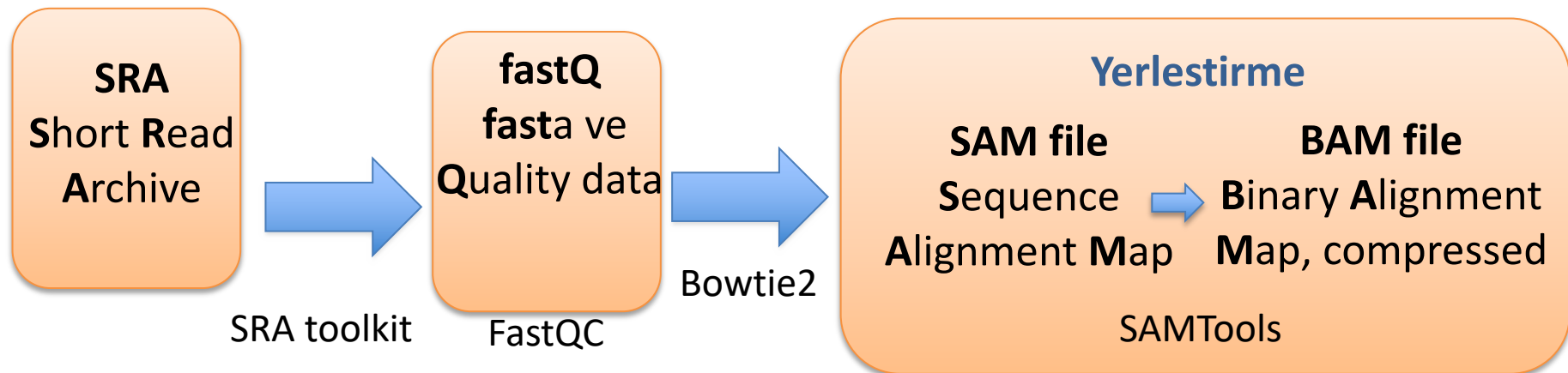
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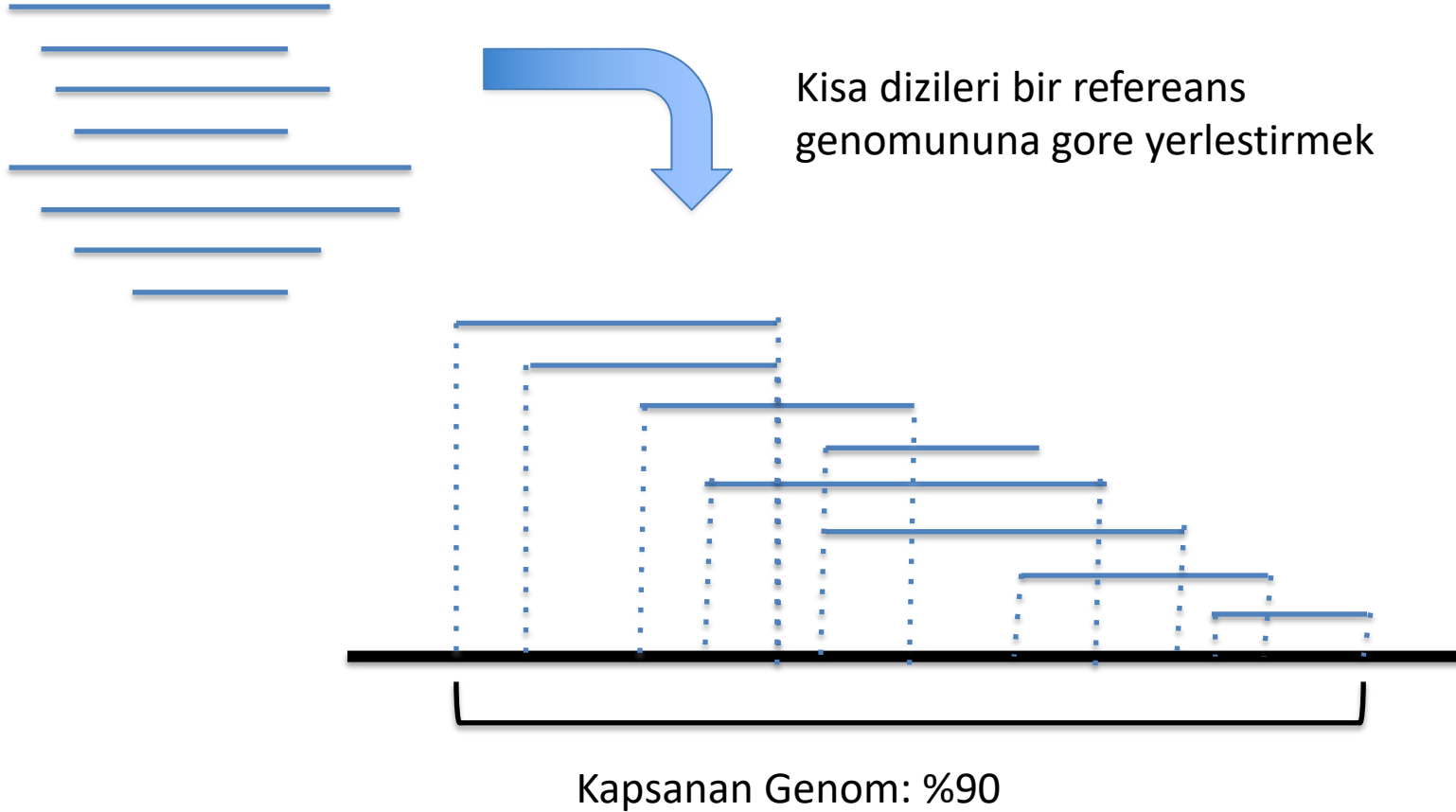
Yerlestime Kalitesi

Bu parçacık genoma yerleştirilirken ne kadar kesin konuşabiliriz?

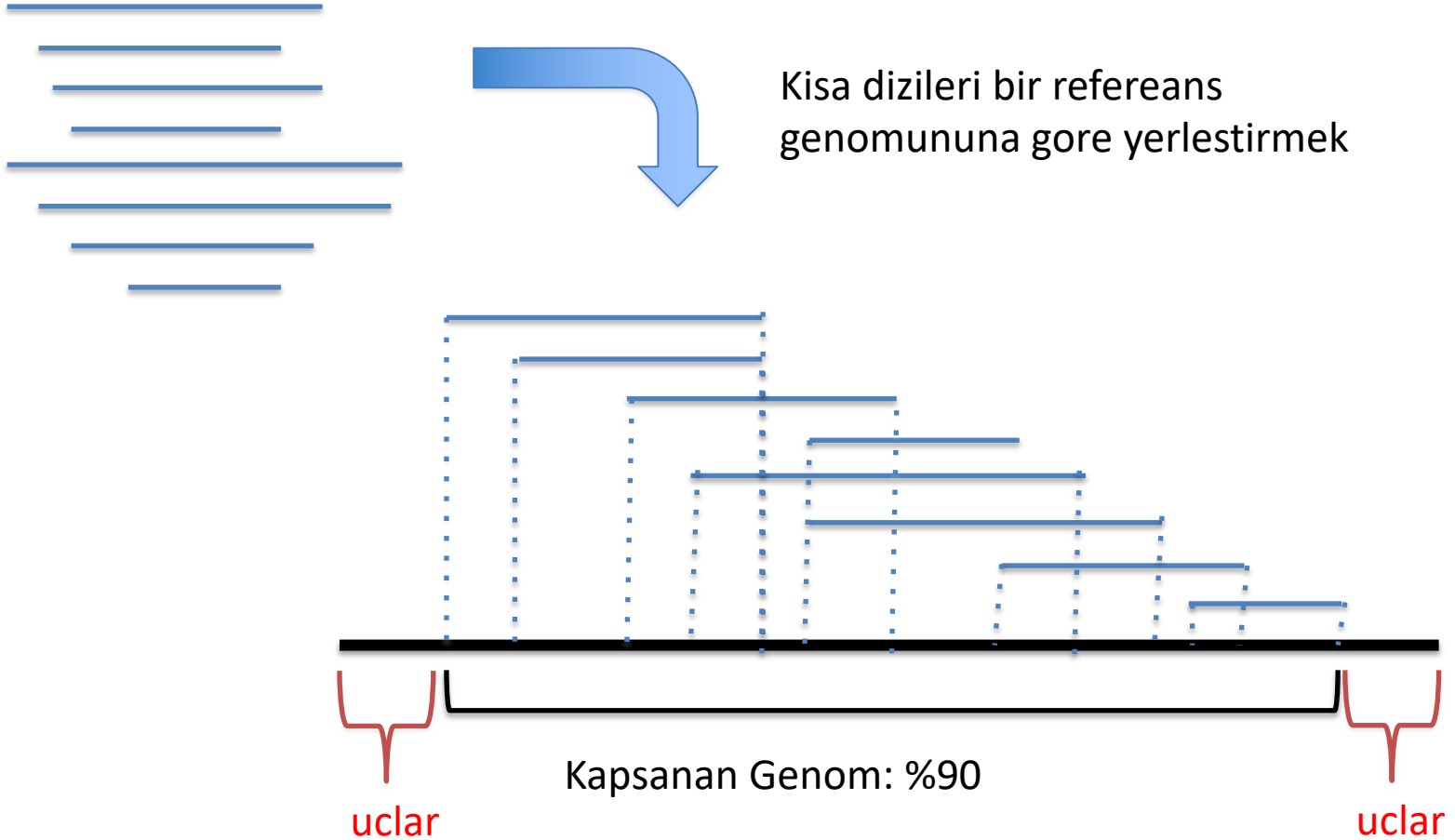




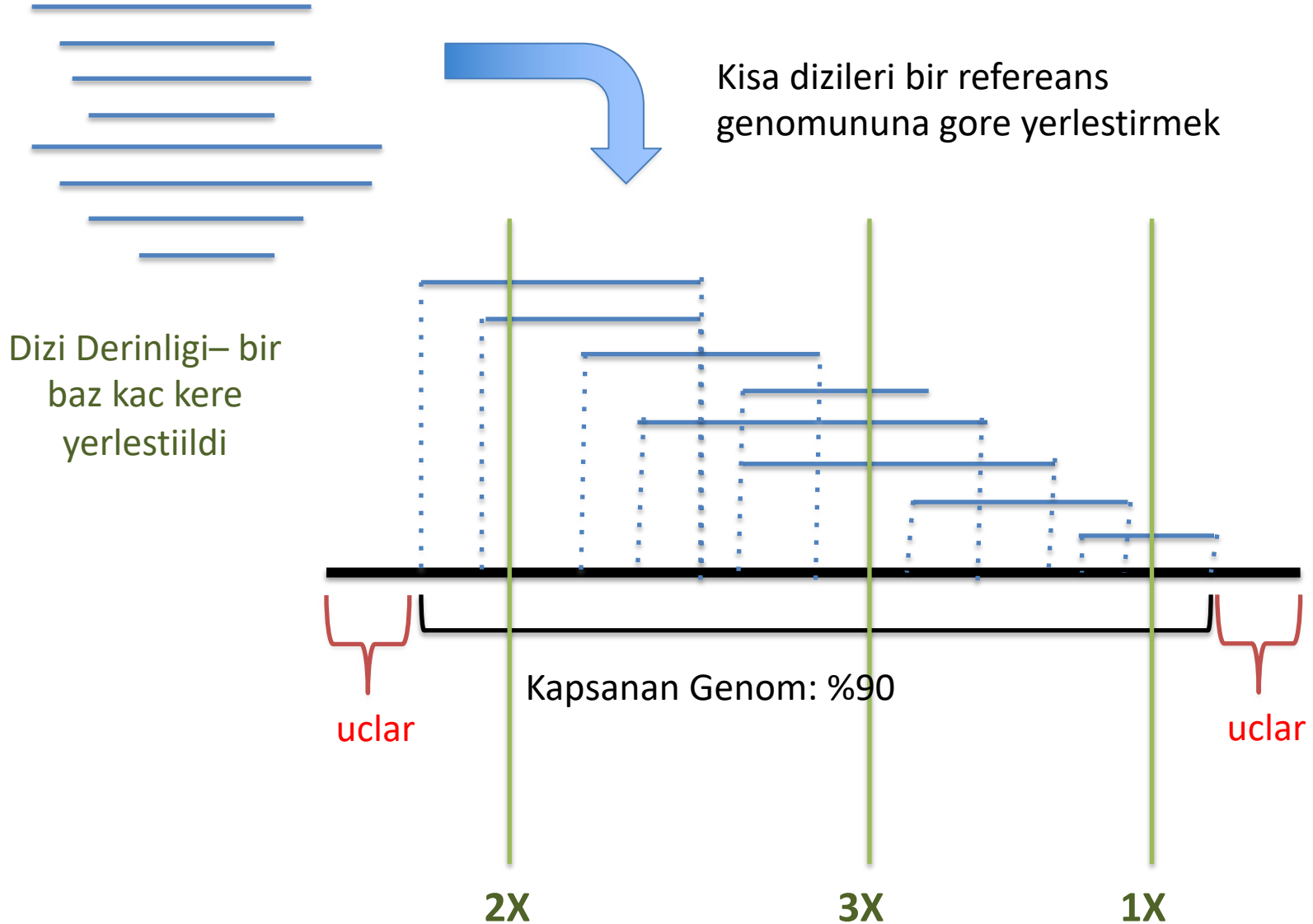
Parcacik Yerlestirme



Parcacik Yerlestirme



Parcacik Yerlestirme

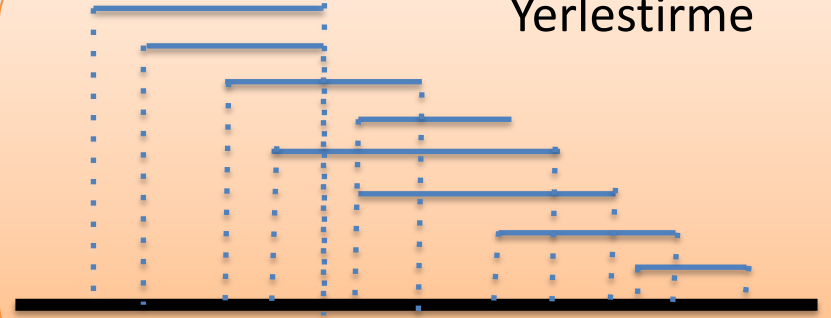




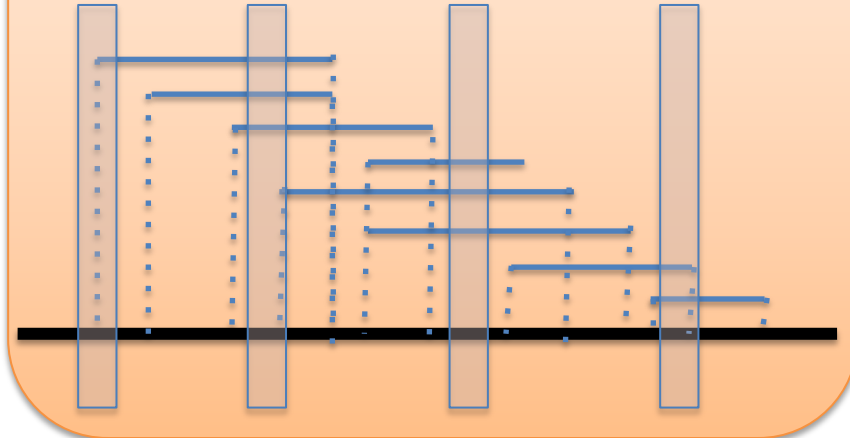
Kisa Diziler



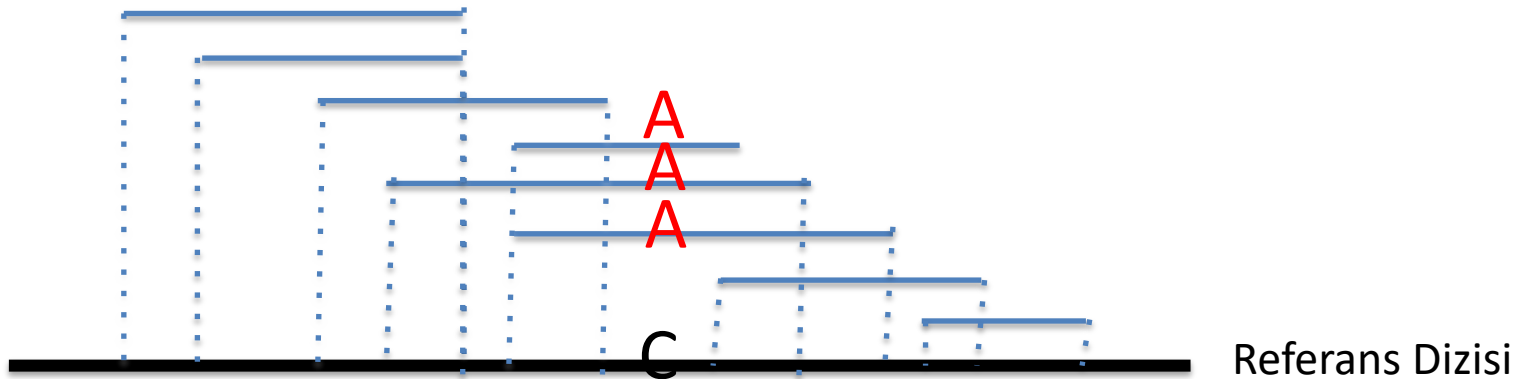
Yerlestirme



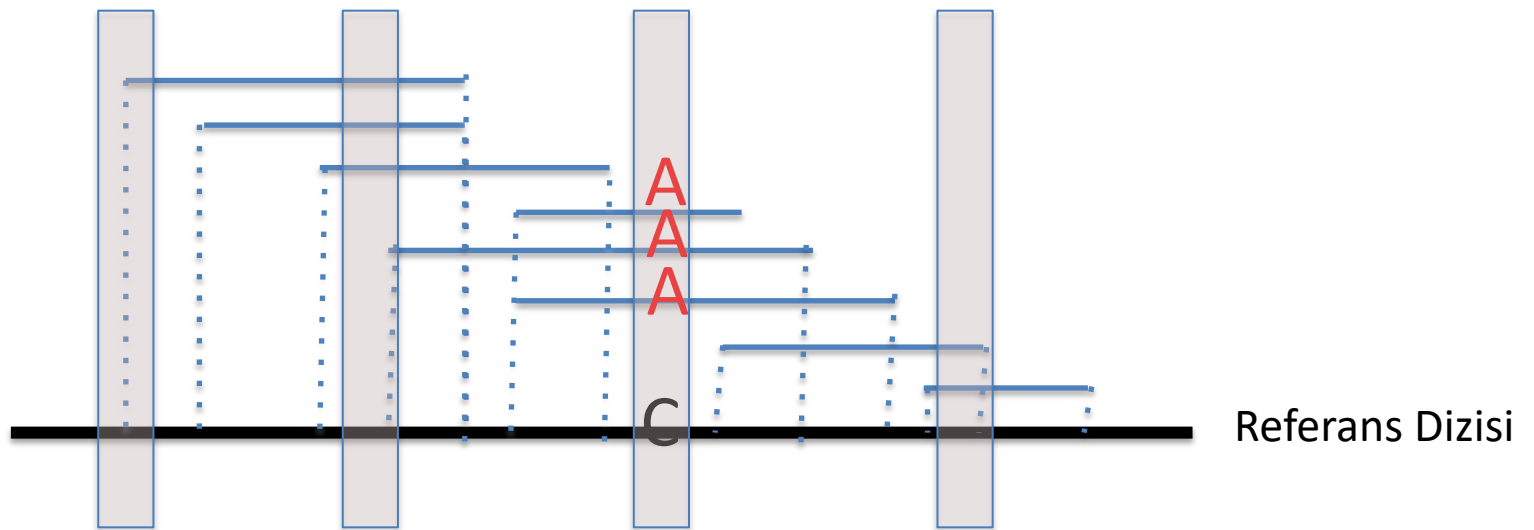
Varyant Tespiti

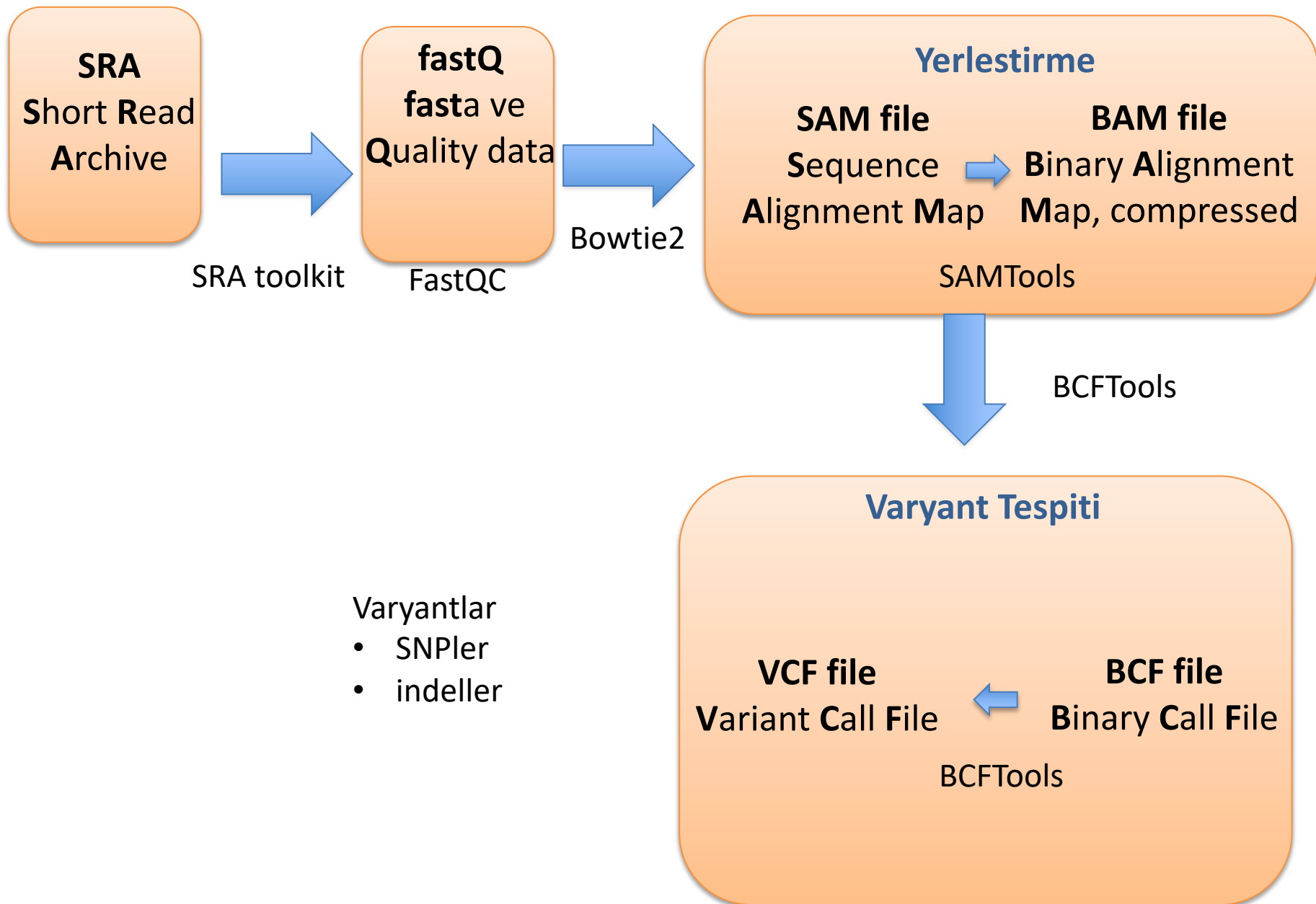


Varyant Tespiti



Varyant Tespiti





Variant Call File Format

Reference/
Alternate Allele

Upstream
pipeline Result

Genotype

##fileformat=VCFv4.0

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chr1	888659	3748597	T	C	28	PASS	DP=7	GT:DP	1/1:7
chr1	900286	.	AG	A	3	FAIL	DP=1	GT:DP	1/1:1
chr1	900505	28705211	G	C	0	PASS	DP=5	GT:DP	0/1:5
chr1	908823	28687780	G	A	29	PASS	DP=18	GT:DP	1/0:18
chr1	948921	15842	T	C	.	PASS	DP=28	GT:DP	1/.:28
chr1	949608	1921	G	A	44	PASS	DP=7	GT:DP	0/1:7
chr1	949654	8997	A	G	31	PASS	DP=16	GT:DP	1/1:16
chr1	980460	.	G	A	2	FAIL	DP=2	GT:DP	./.:2
chr1	982941	3128102	T	C	8	PASS	DP=15	GT:DP	1/1:15
chr1	1158631	.	AG	CC	75	PASS	DP=21	GT:DP	1/1:21
chr1	1225612	.	G	A	5	FAIL	DP=4	GT:DP	1/1:4
chr1	1226063	.	G	A	26	PASS	DP=6	GT:DP	0/1:6

Chromosome

Position

Call Quality

Read Depth either
in INFO or within
each samples