

Dizi Yerleştirme ve Varyant Tespiti Pratigi

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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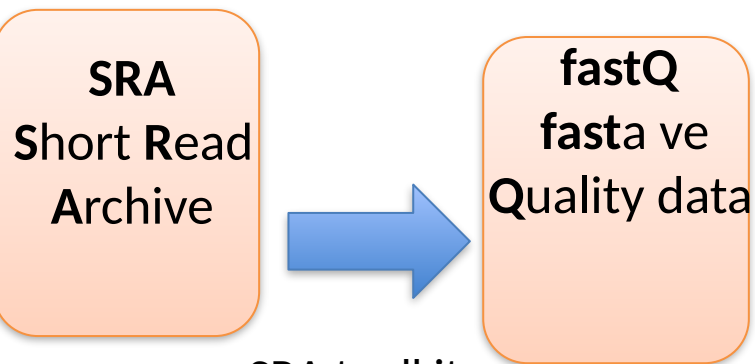
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<< First < Prev Page of 8 [Next >](#) [Last >>](#)

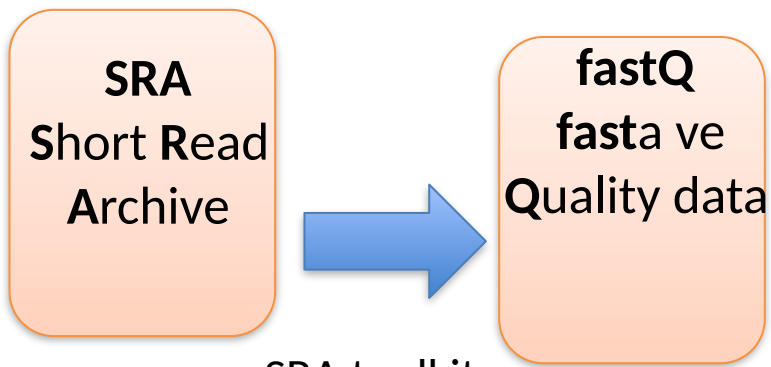
 Showing SRA Experiments for SRP047378

- ☐ [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

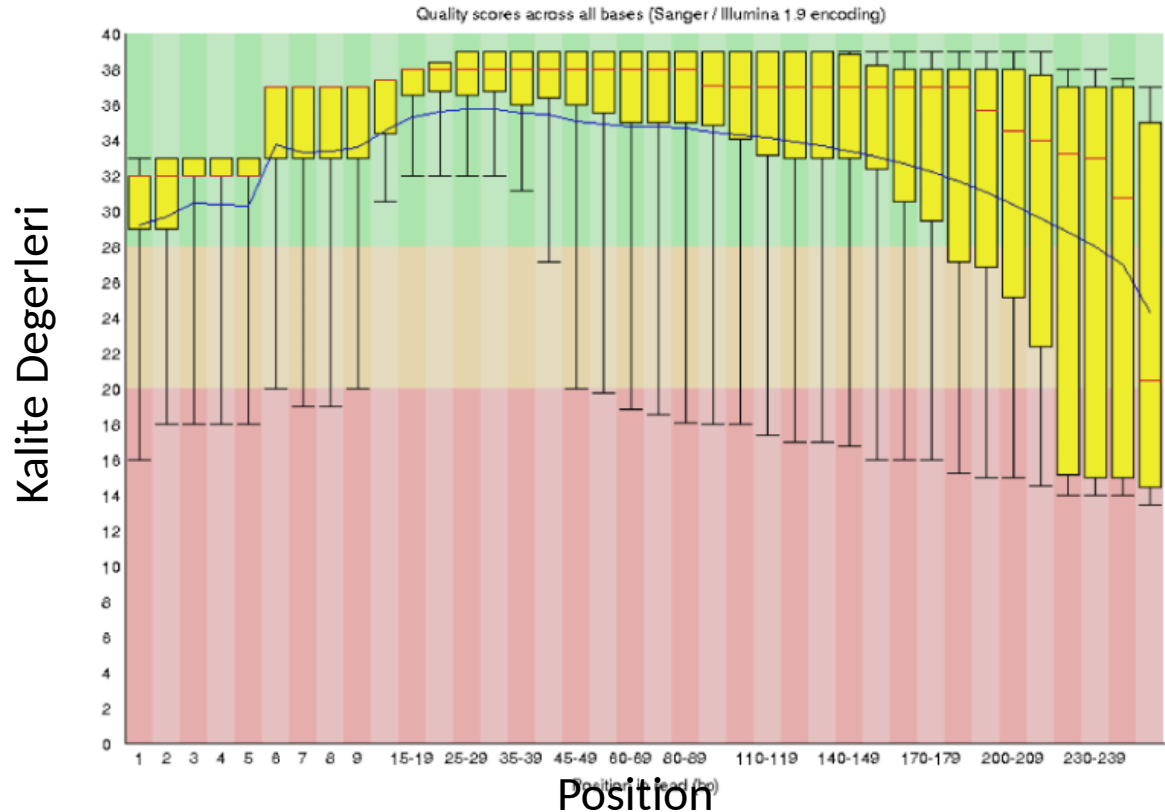


- Gene ID, uzunluk
- Fasta dizisi
- Gene ID, uzunluk
- Kalite verisi

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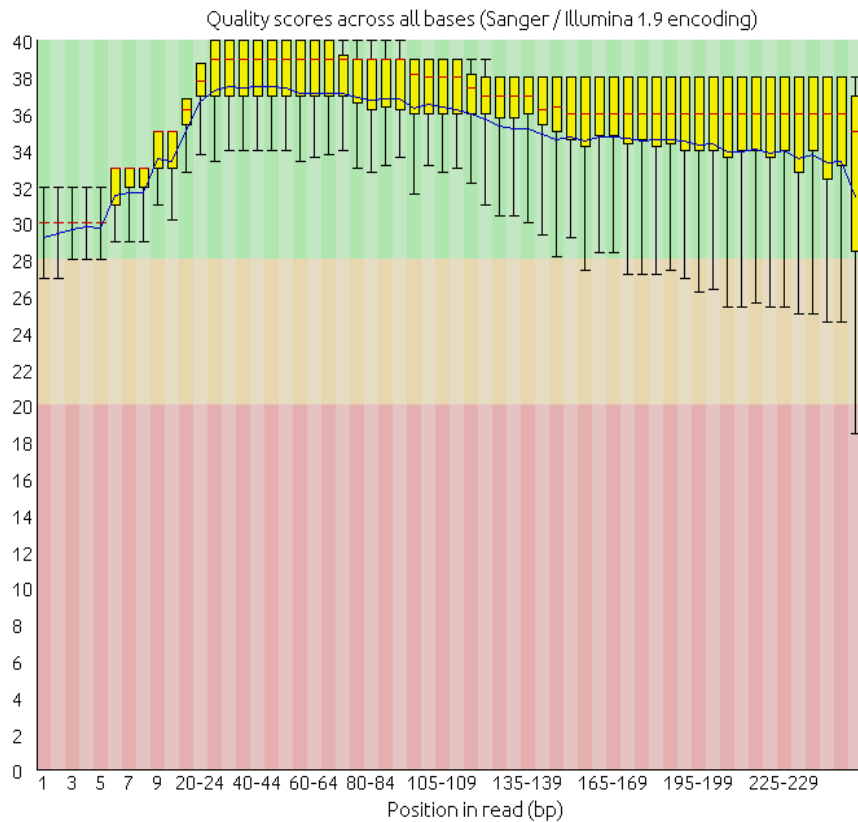


FastQC

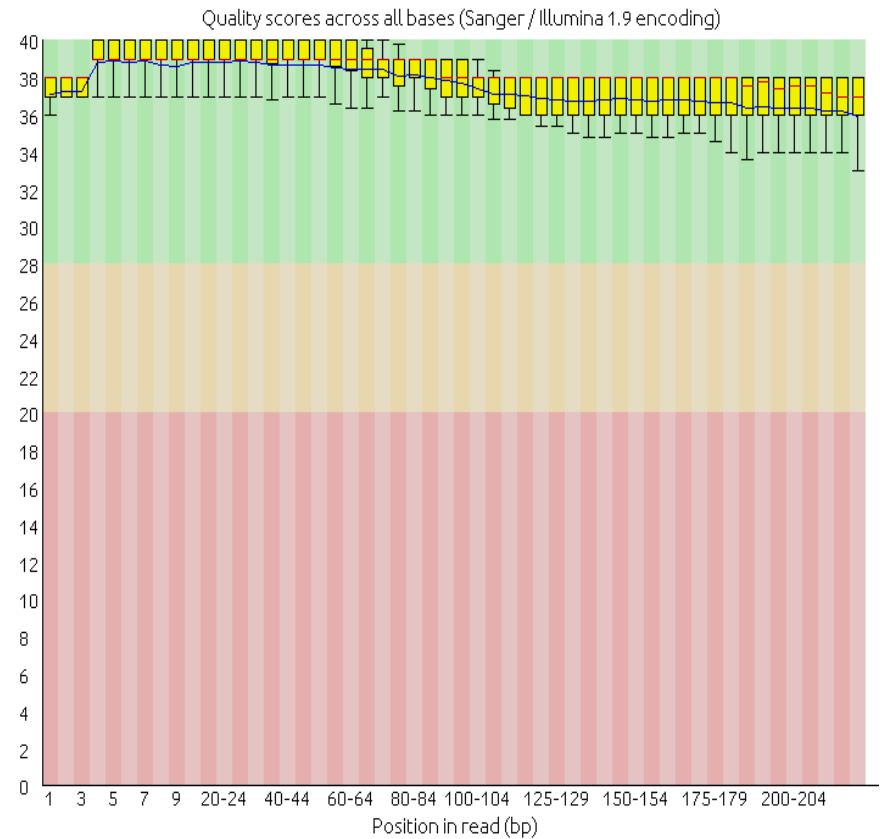


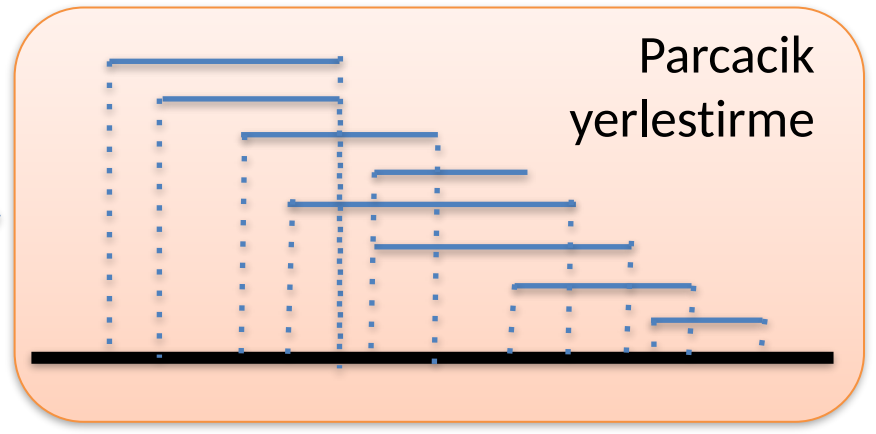
- Bastaki ve sondaki kotu kalite bazlari kes
- Aralardaki kotu kalite bazlari filtrele

Oncesi

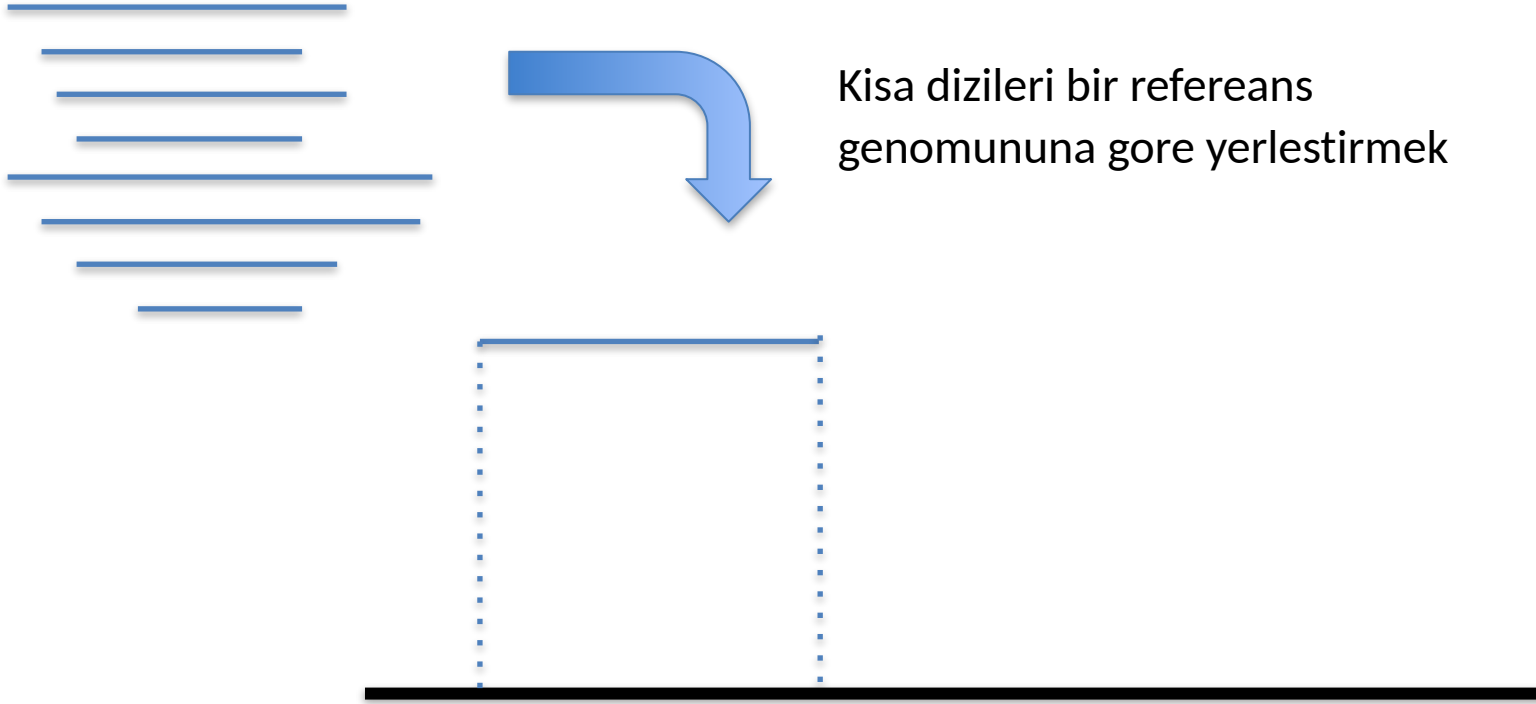


Sonrasi

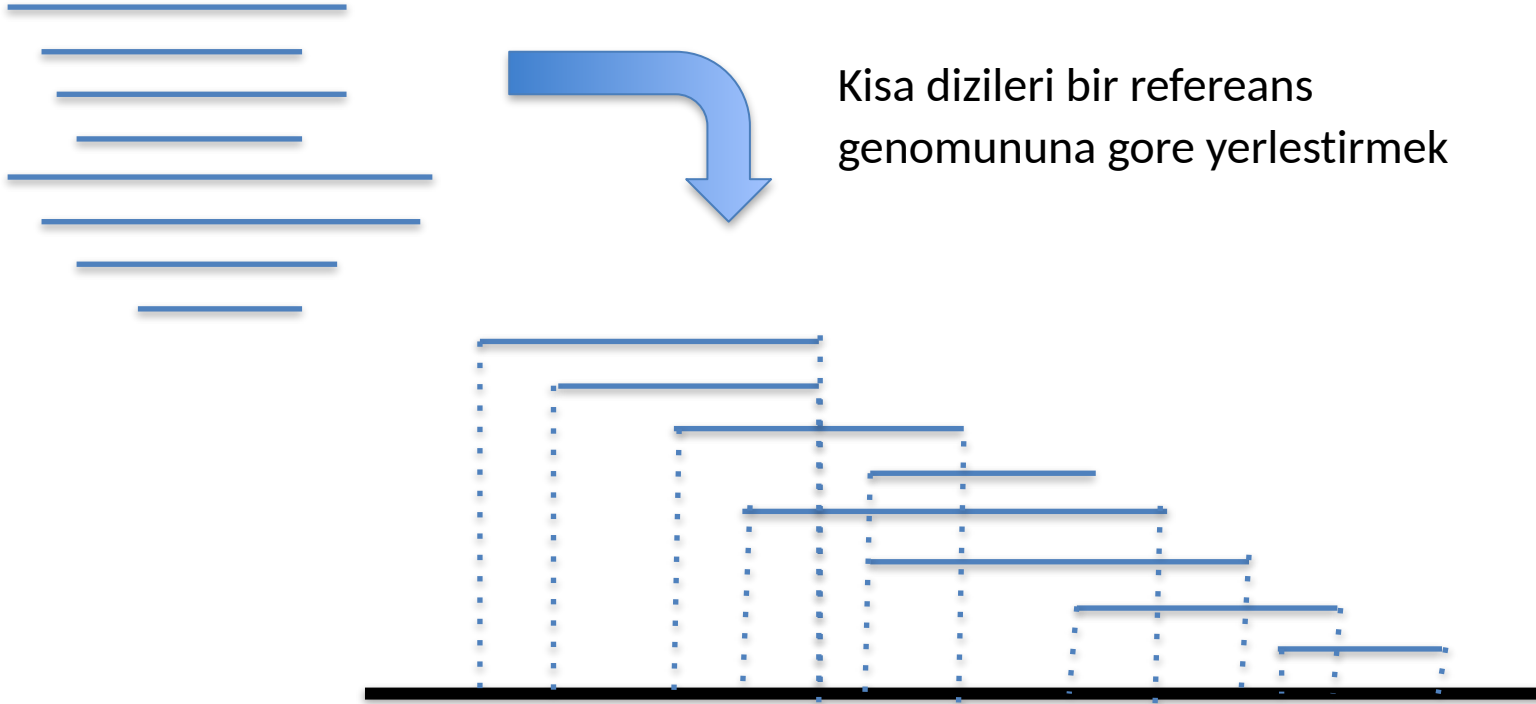




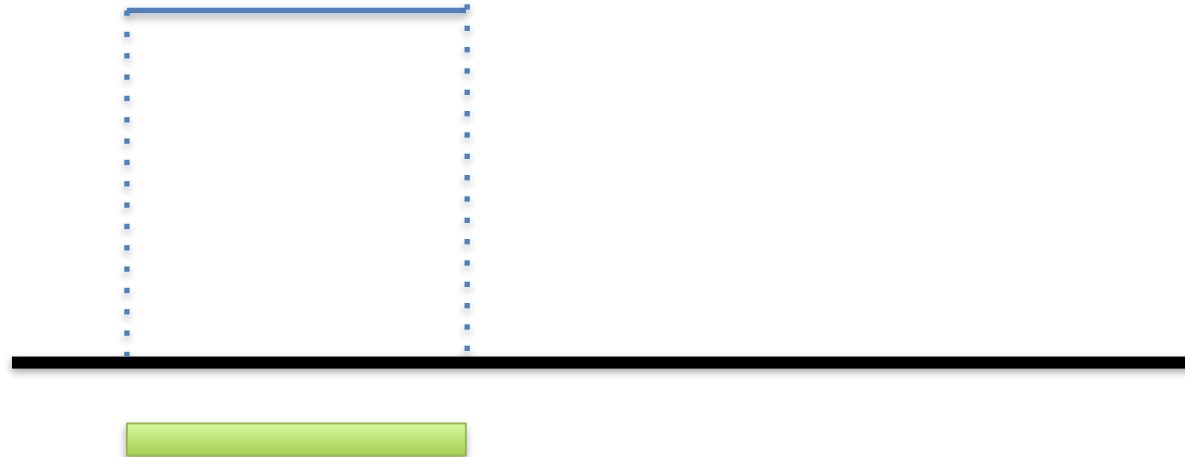
Parcacik Yerlestirme



Parcacik Yerlestirme



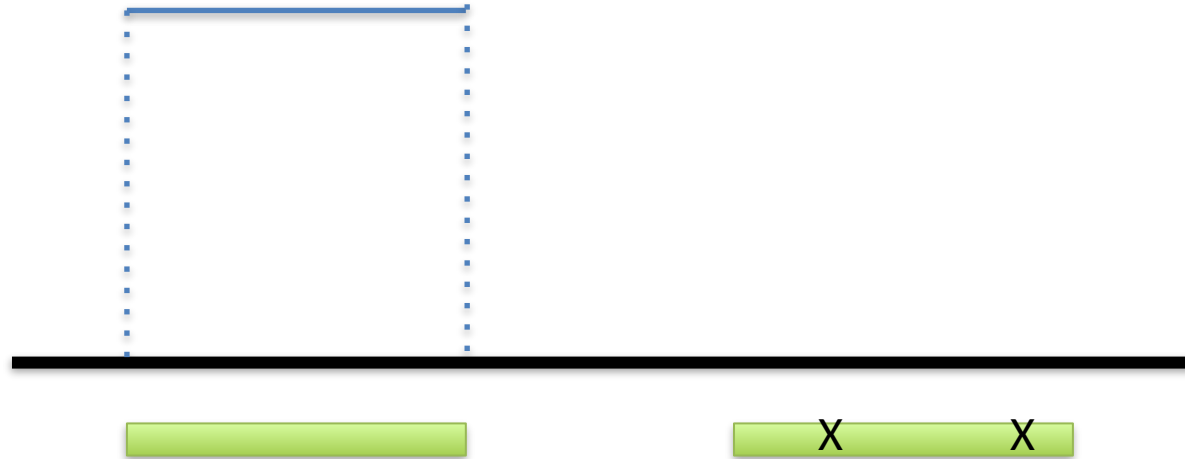
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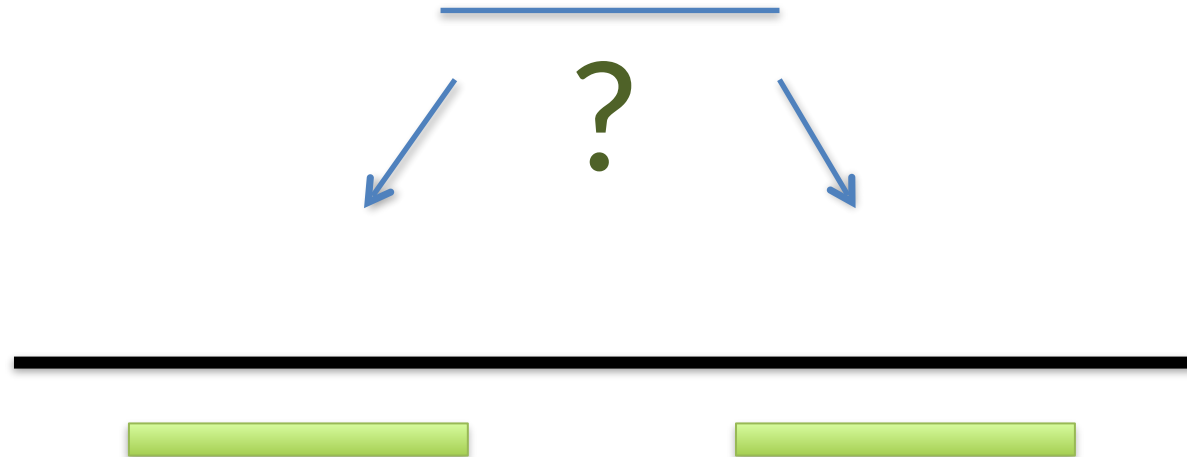
Yerleştirmeye Kalitesi



Yerleştirmeye Kalitesi

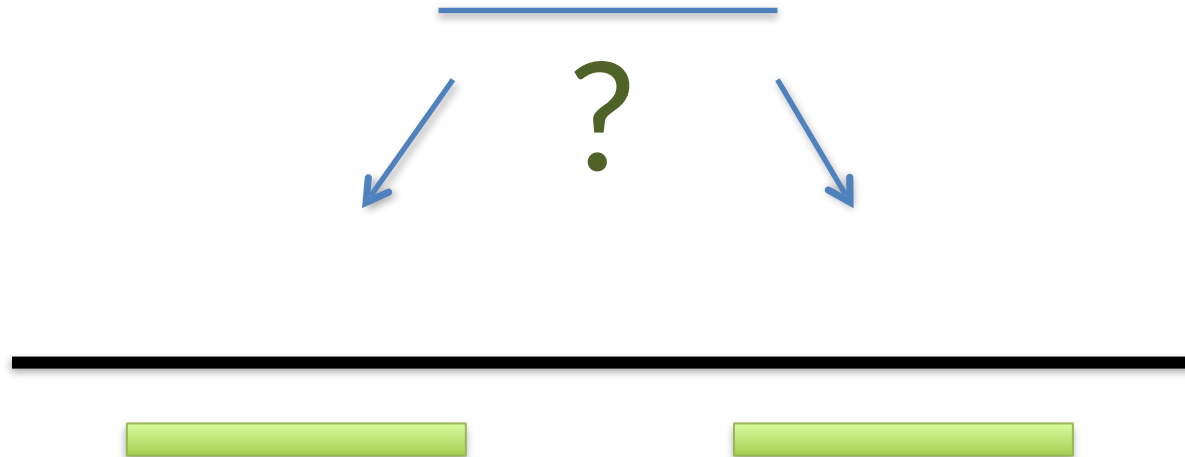


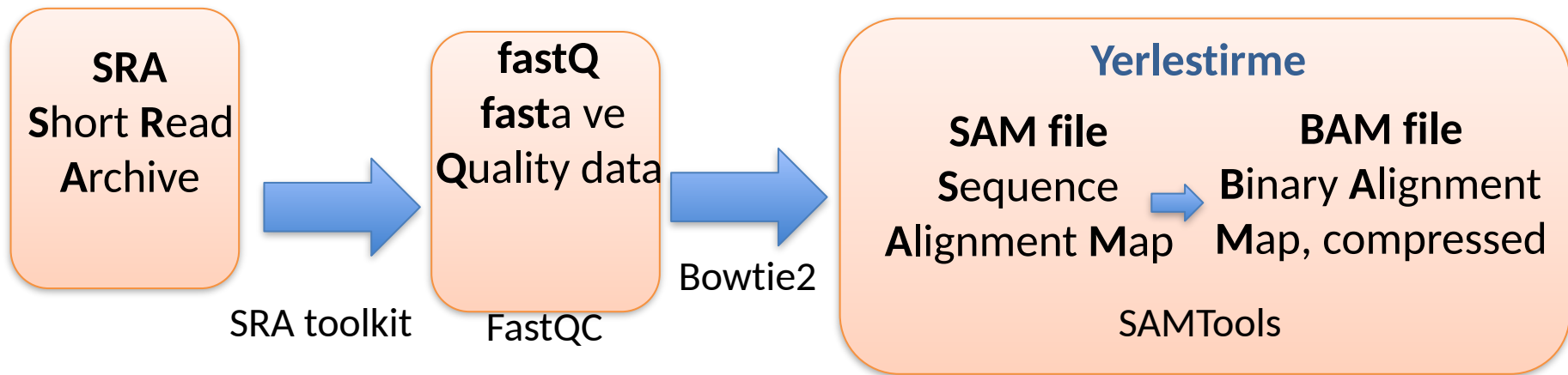
Yerleştirmeye Kalitesi



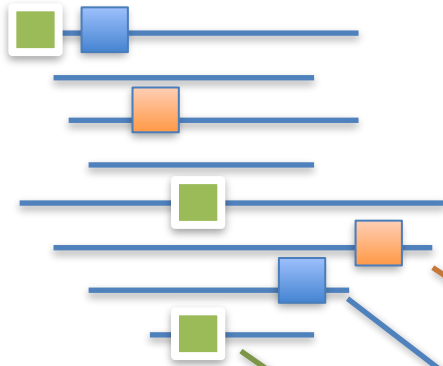
Yerleştirmeye Kalitesi

Bu parçacık genomaya yerleştirilirken ne kadar kesin konuşabiliriz?





Indexleme

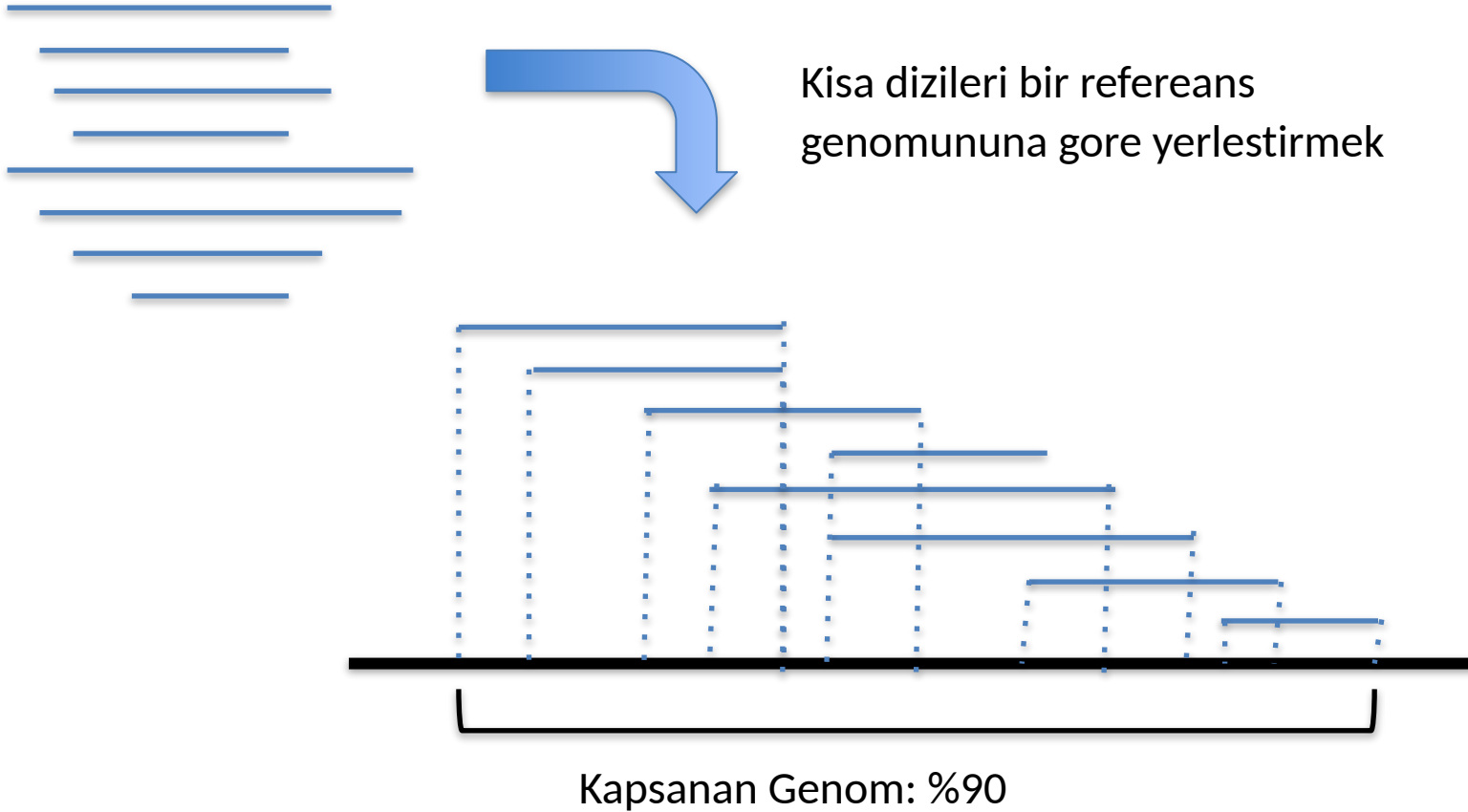


Her işlem öncesinde kullandığımız aracın kendi indeksleme komutuyla dizilerimizi indeksliyoruz.

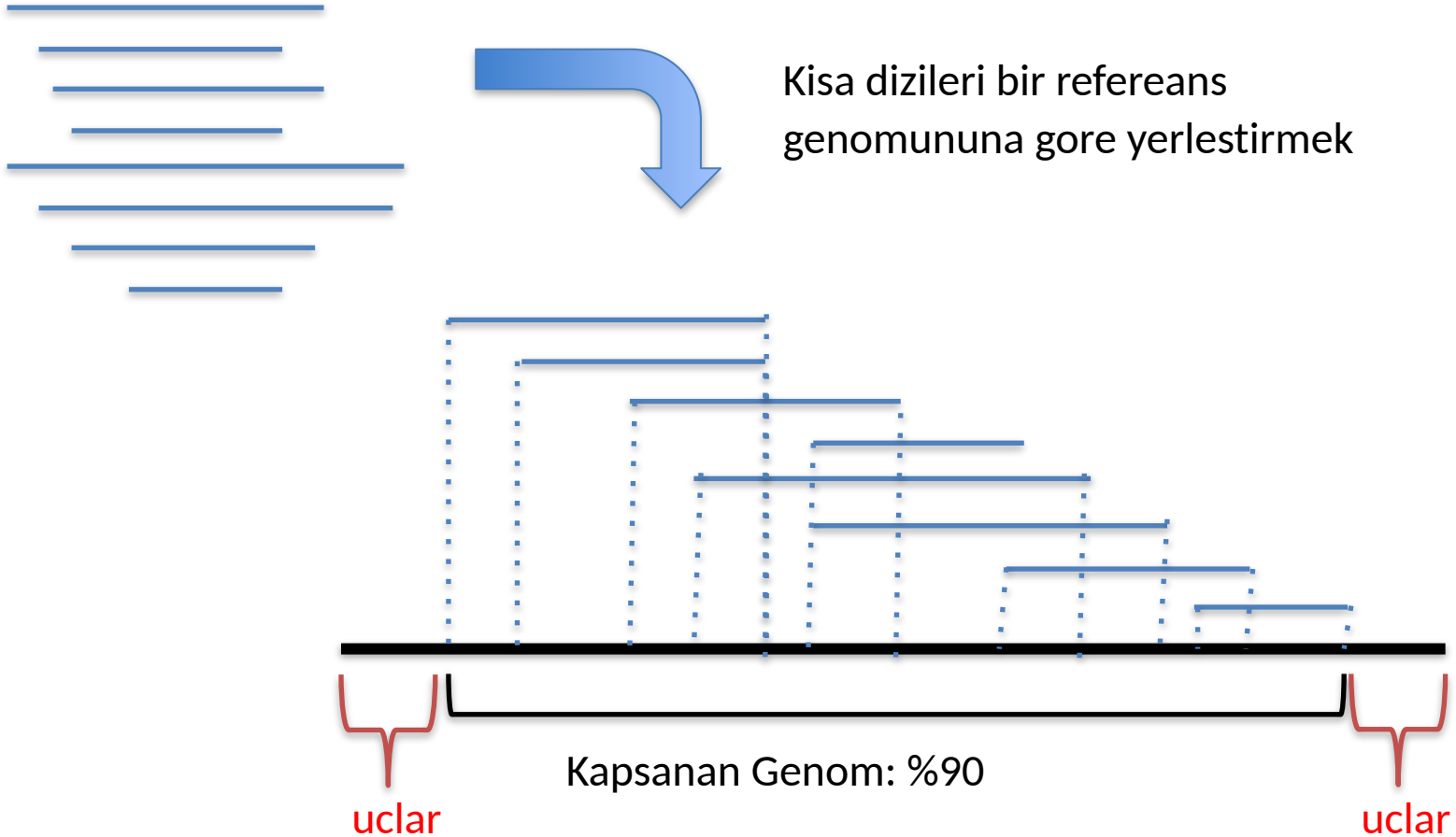
Kök	Pozisyon
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ACCT	4,16,24
GTGA	12,20
...	...

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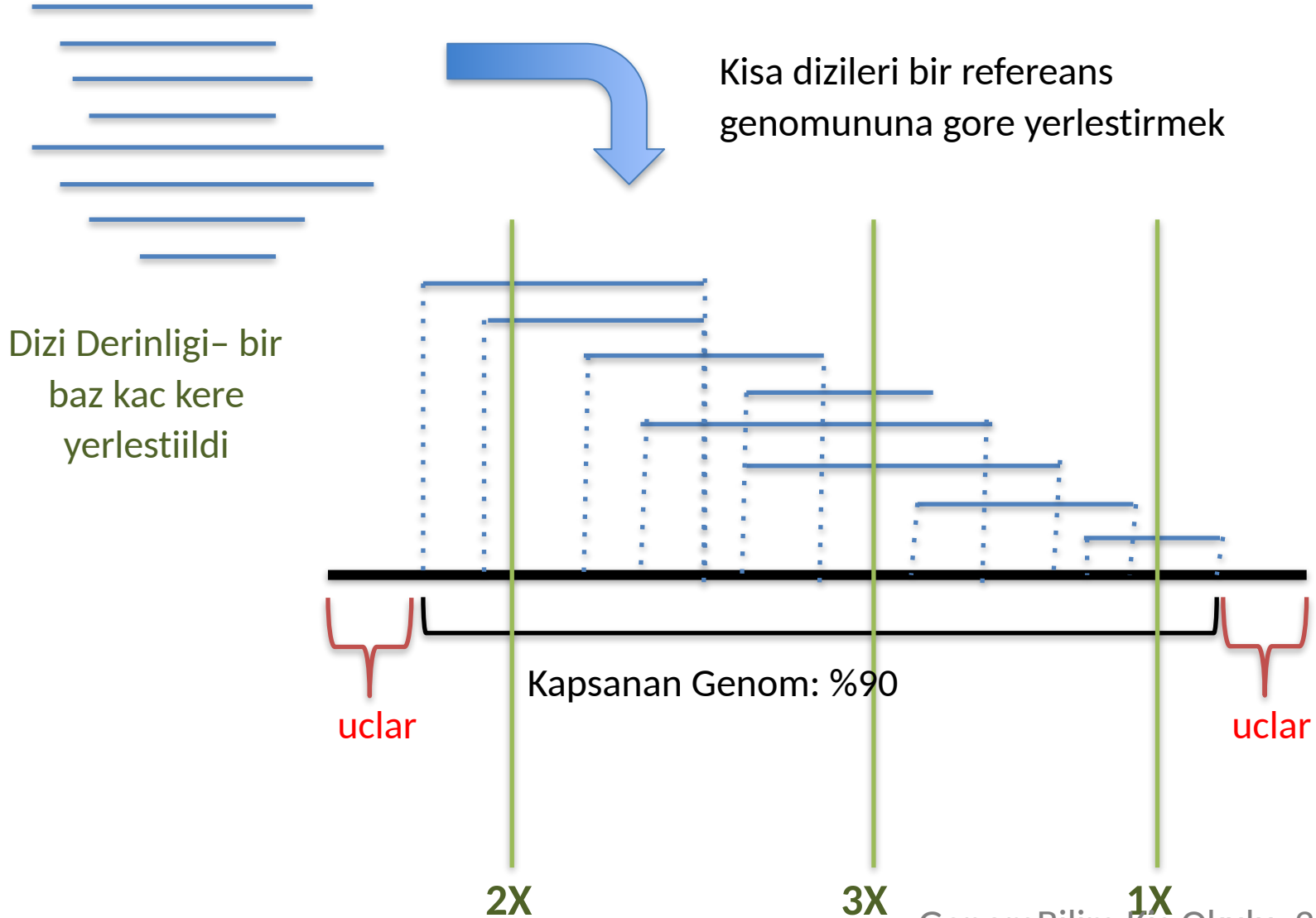
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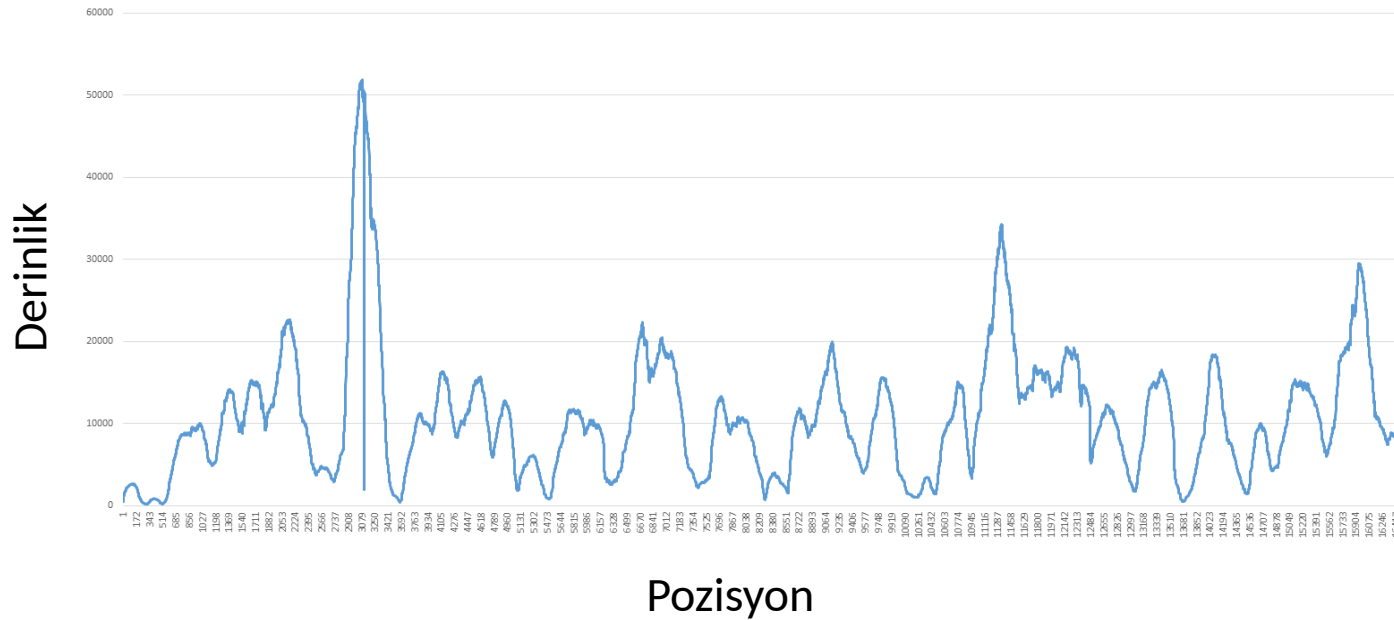
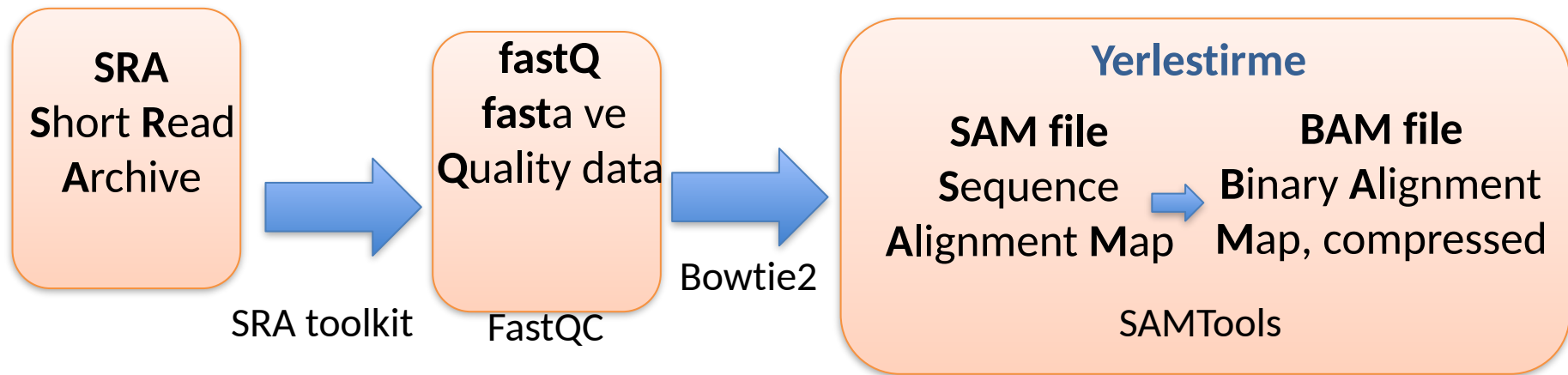


Parcacik Yerlestirme

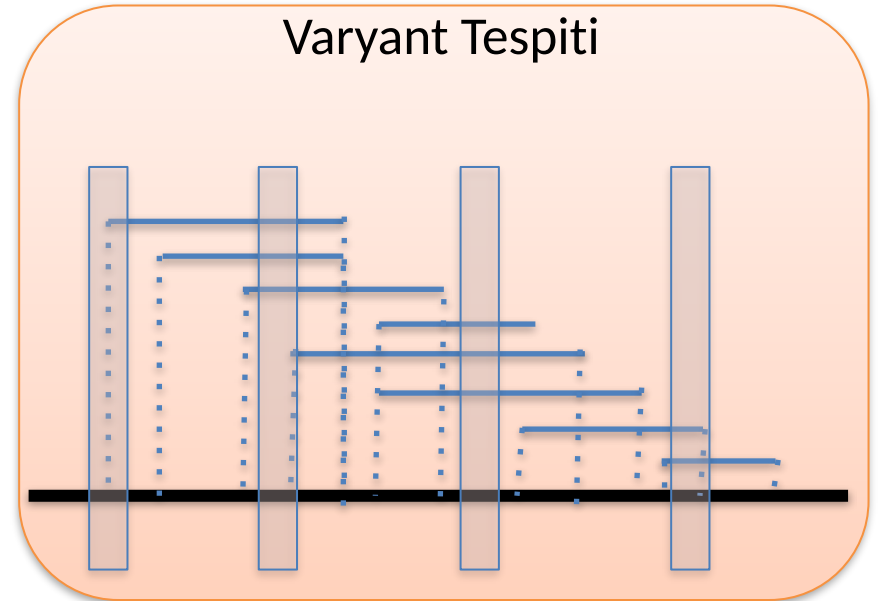
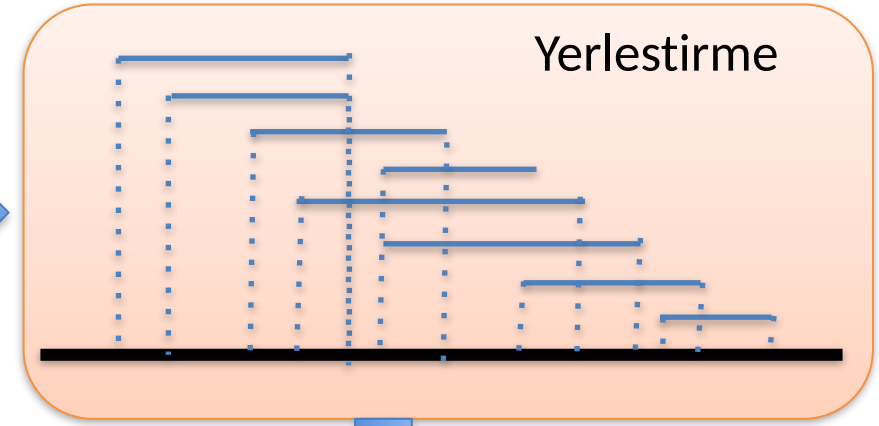


Parcacik Yerlestirme

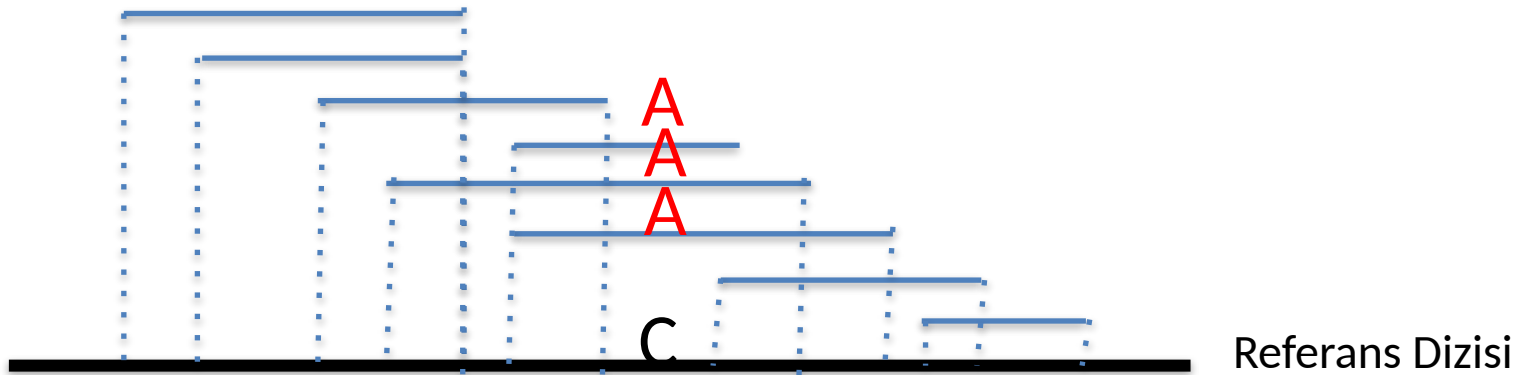




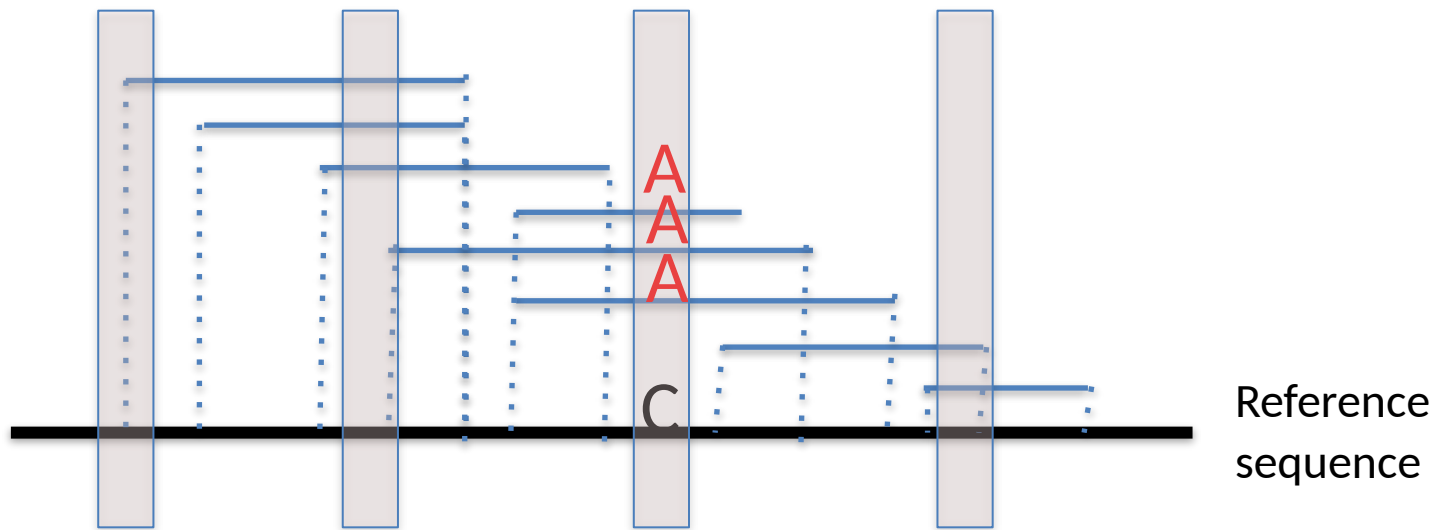
- Kapsam
- Derinlik

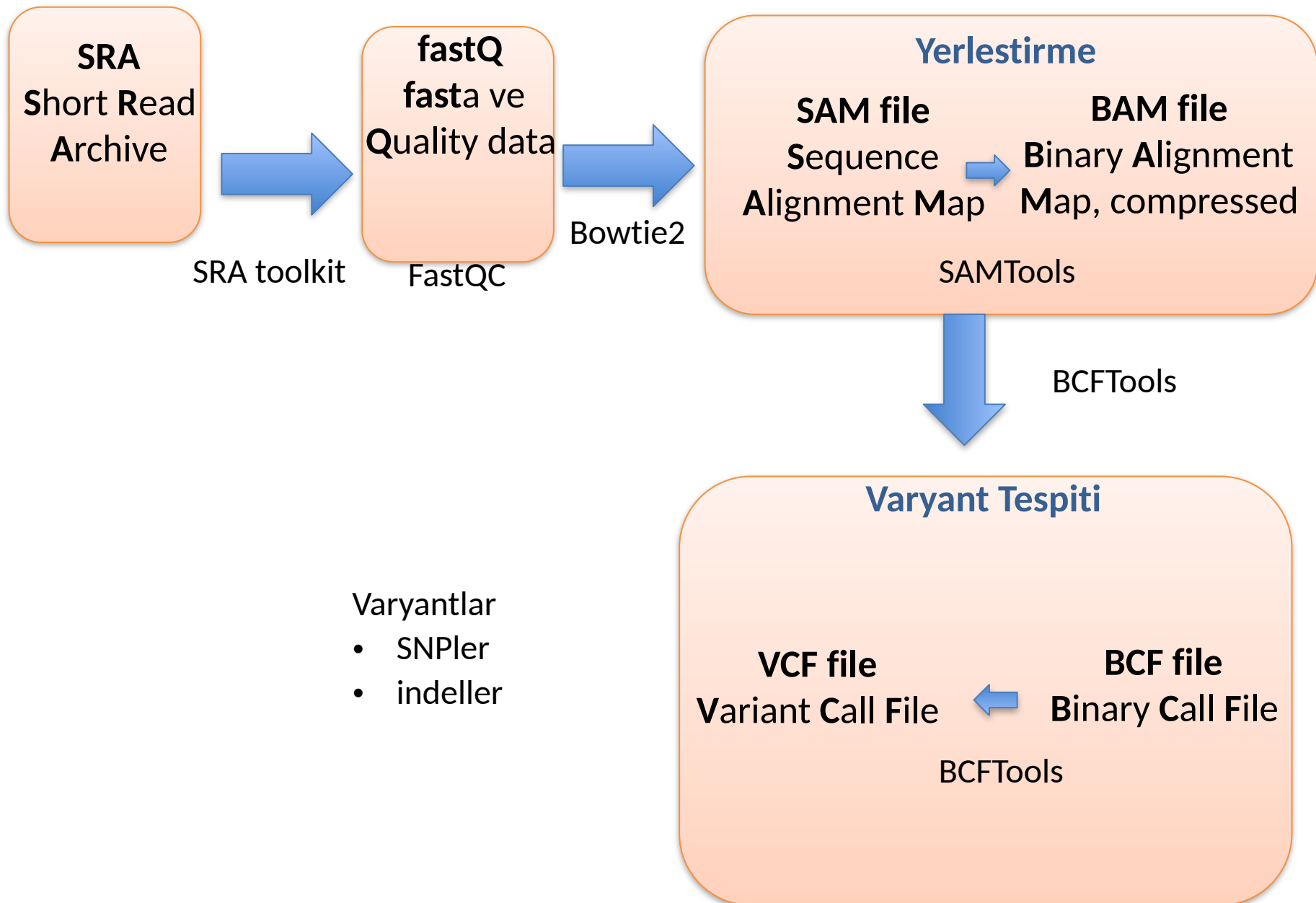


Varyant Tespiti



Varyant Tespiti





Variant Call File Format

Reference/
Alternate Allele

Upstream
pipeline Result

Genotype

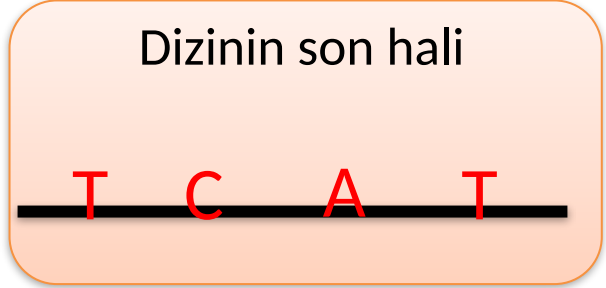
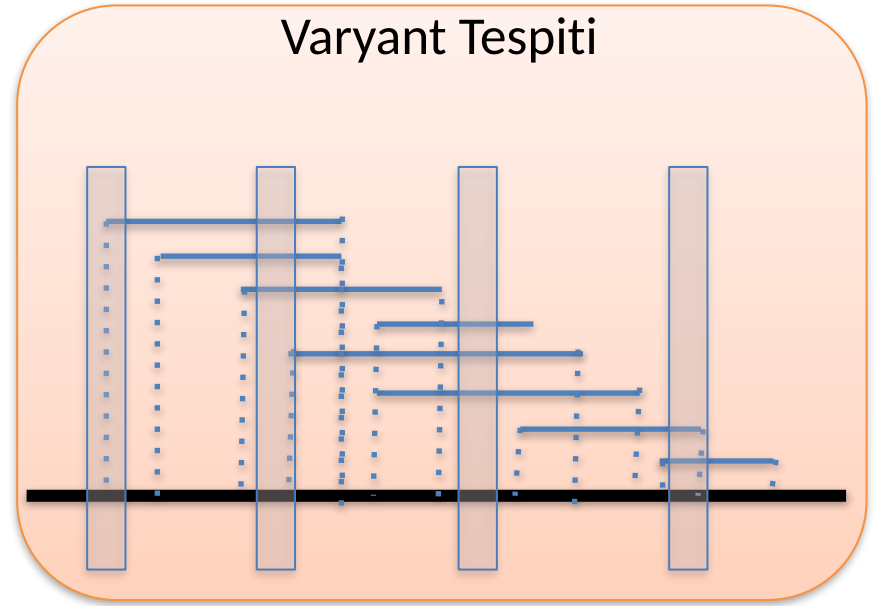
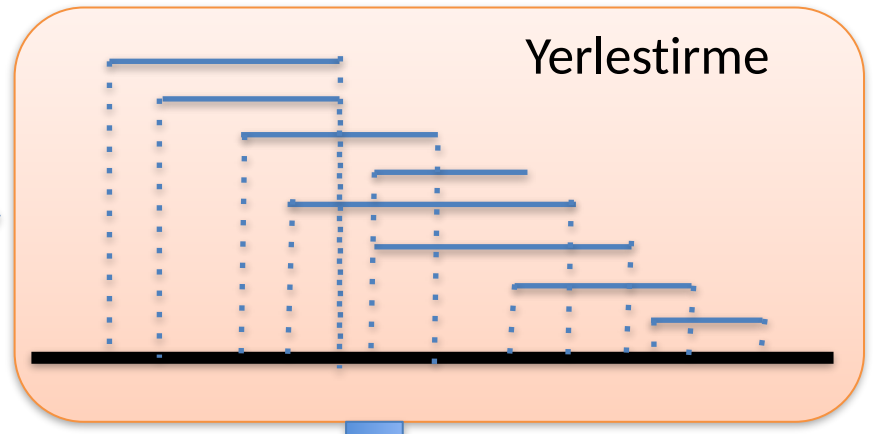
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chr1	888659	3748597	T	C	28	PASS	DP=7	GT:DP	1/1:7
chr1	900286	.	AG	A	3	FAIL	DP=1	GT:DP	1/1:1
chr1	900505	28705211	G	C	0	PASS	DP=5	GT:DP	0/1:5
chr1	908823	28687780	G	A	29	PASS	DP=18	GT:DP	1/0:18
chr1	948921	15842	T	C	.	PASS	DP=28	GT:DP	1/.:28
chr1	949608	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
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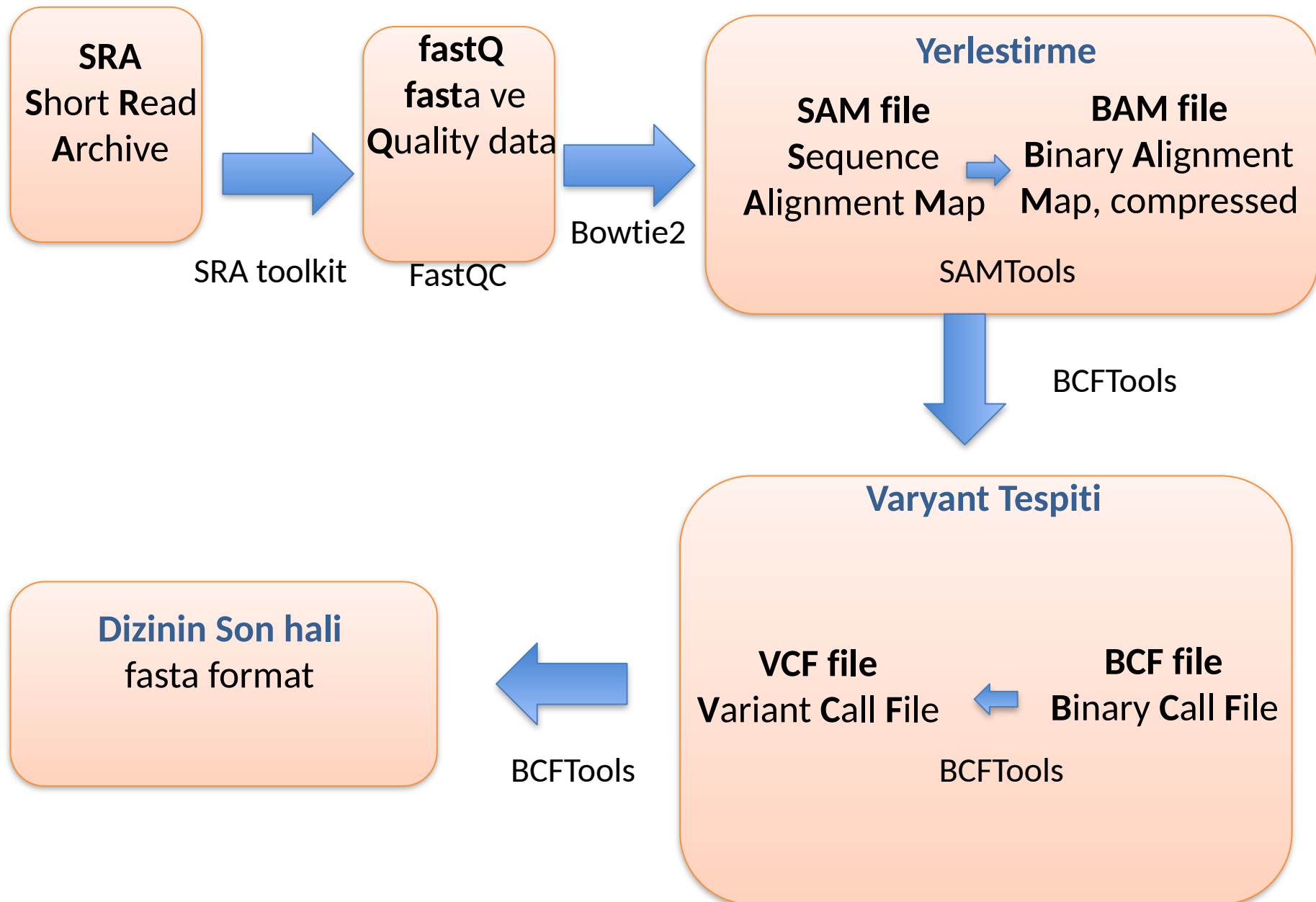
Chromosome

Position

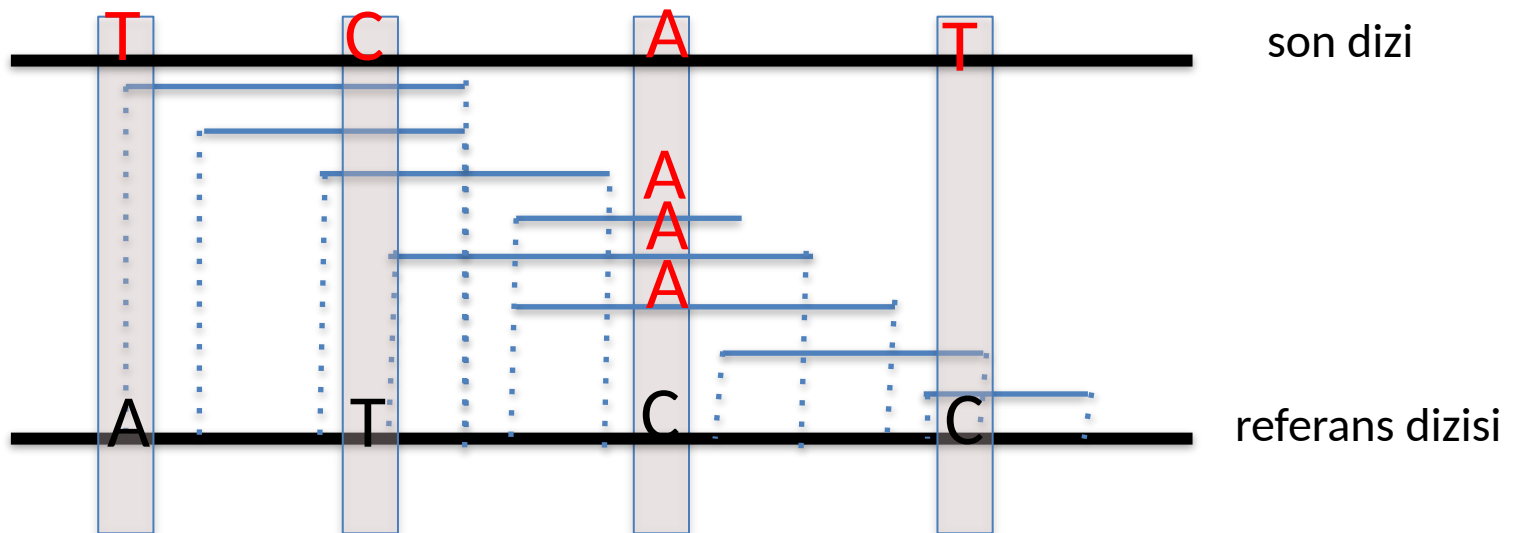
Call Quality

Read Depth either in INFO or within each samples





Dizinin Son Hali



Dizinin Son Hali

