GTrack 1.0:

Unified data format providing customizable representation and high-speed analysis performance within Galaxy

Sveinung Gundersen, PhD student
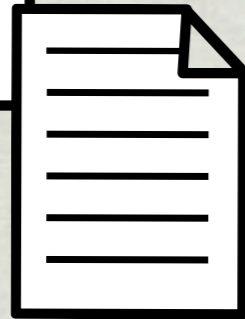The Norwegian Radium Hospital, OUH

September 3rd, 1967, Stockholm, Sweden

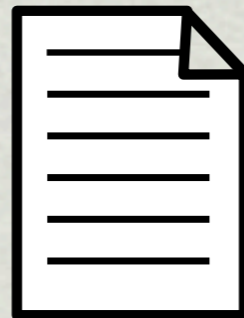The day Sweden changed from driving on the left to driving on the right
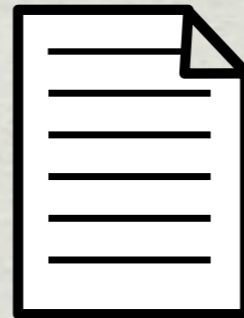
File formats for genomic track data
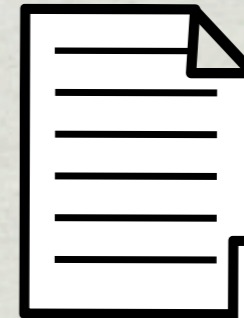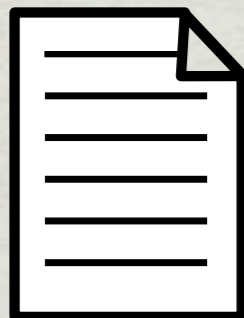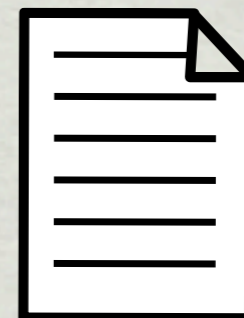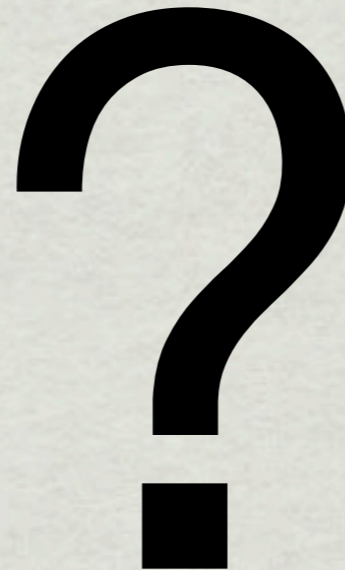
Why couldn't our world just be like this?
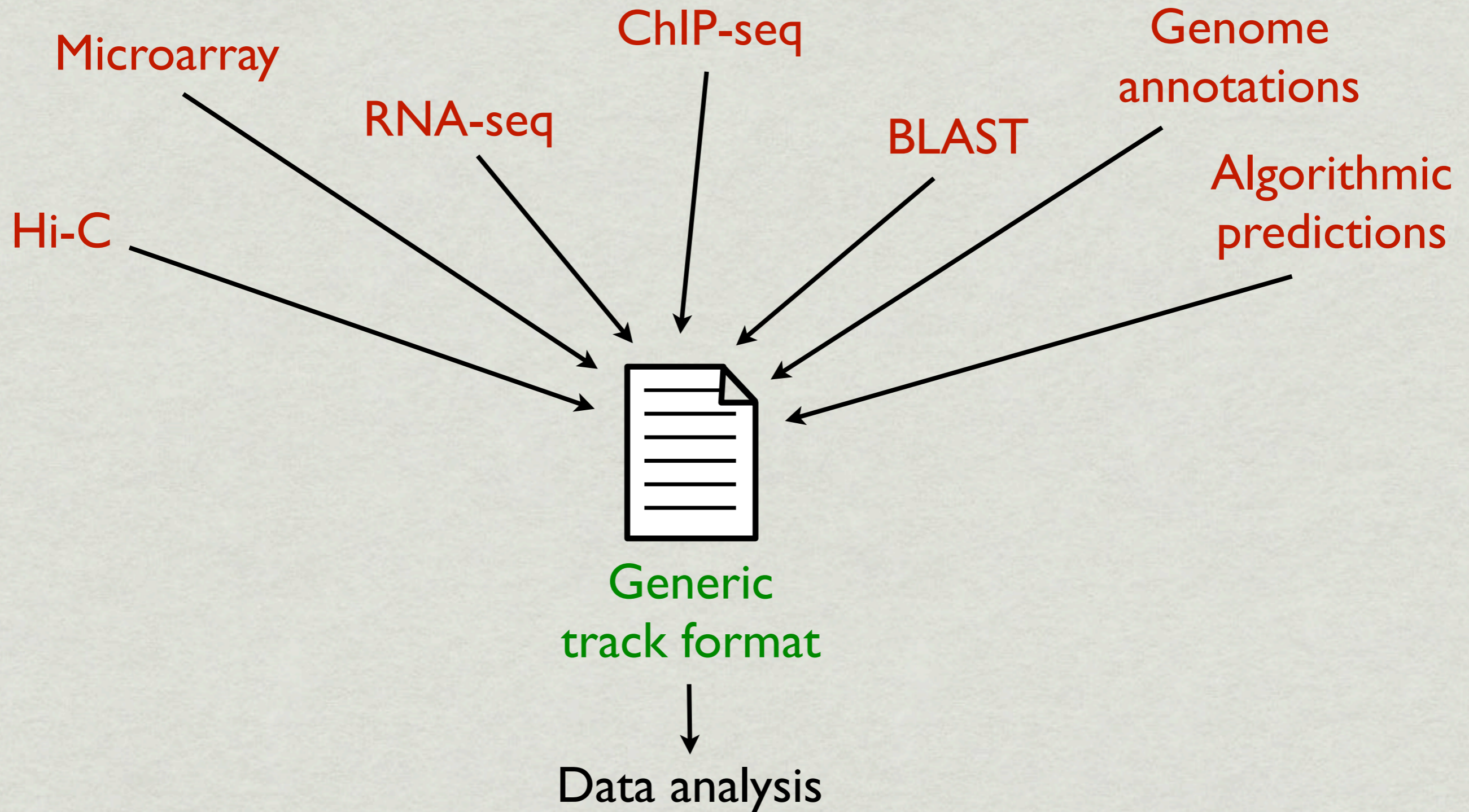
Why all the formats?

1. Different columns

2. Underlying structural differences

3. Format-specific characteristics

4. Tool-specific format requirements

5. Performance issues

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Different columns

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- Different set of columns in the different formats

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- Solution: handle flexible columns in one format

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- Galaxy interval format handles flexible columns:

```
#CHROM    START    END      STRAND    NAME    COMMENT
chr1      10       100      +         exon    myExon
chrX      1000     10050    –         gene    myGene
```

Different columns

- Different set of columns in the different formats

- Solution: handle flexible columns in one format

- Galaxy interval format handles flexible columns:

```
#CHROM    START    END      STRAND    NAME    COMMENT
chr1      10       100      +         exon    myExon
chrX      1000     10050    –         gene    myGene
```

- However:

  - columns are specified as Galaxy metadata (*i.e.* not readily exportable to other tools)

  - only supports interval data

Different columns

- bigBed also handles flexible columns

- However:

  - columns are specified in server-side AutoSQL files

  - only supports interval data

Why all the formats?

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Underlying data type differences

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- Example: one value per base pair

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- BedGraph format:

```
chr1    0    1      1.0
chr1    1    2     -1.3
chr1    2    3      2.4
...
```

Underlying data type differences

- Example: one value per base pair

- BedGraph format:

```
chr1    0    1      1.0
chr1    1    2     -1.3
chr1    2    3      2.4
...
```

- WIG

```
fixedStep chrom=chr1
1.0
-1.3
2.4

...
```

Underlying data type differences

- Example: one value per base pair

- BedGraph format: Size (Human genome)

```
chr1    0    1      1.0
chr1    1    2     -1.3
chr1    2    3      2.4
...
```
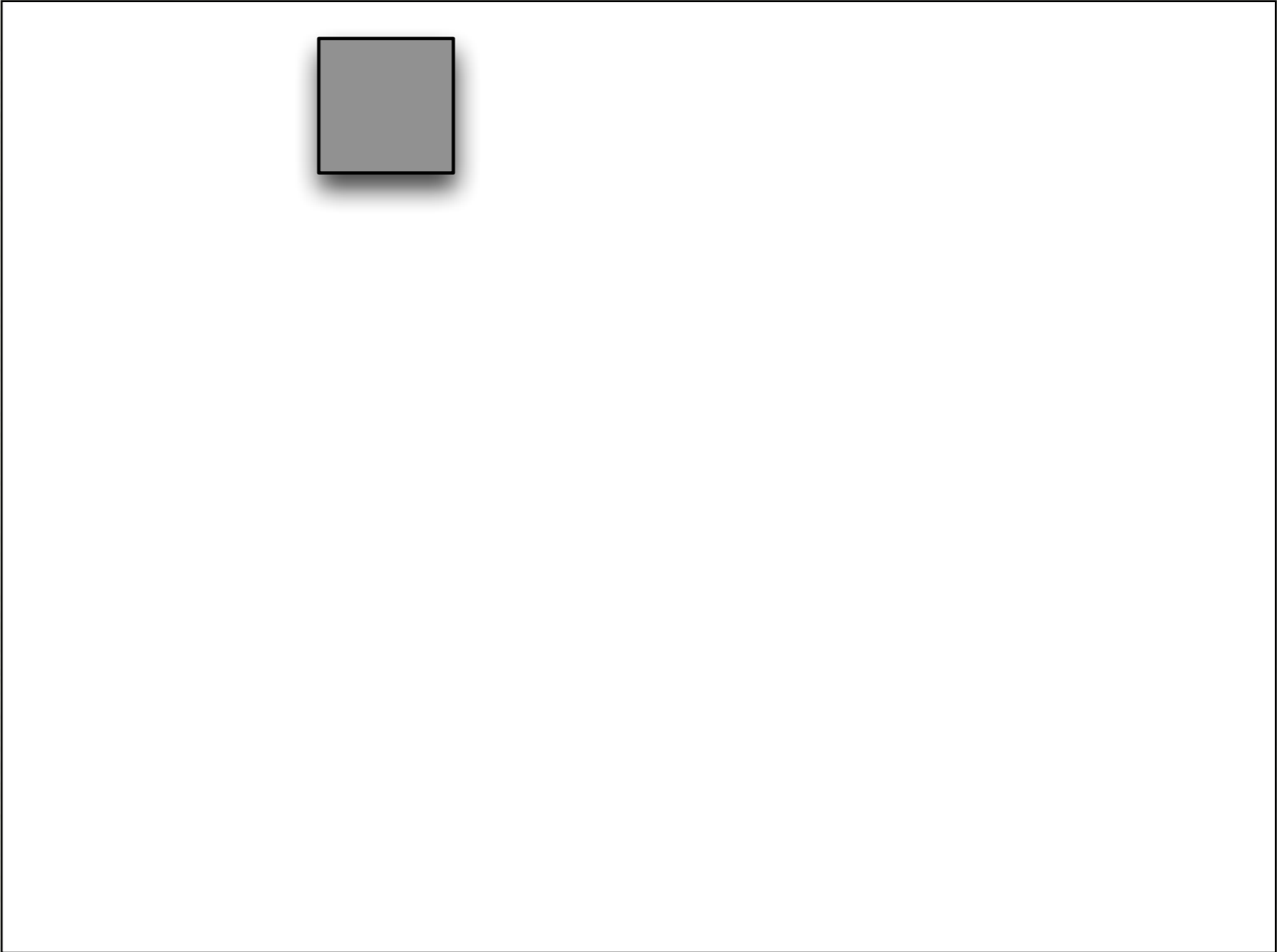
100 GB

- WIG

```
fixedStep chrom=chr1
1.0
-1.3
2.4
...
```

20 GB

No
information

Track
geometry

Genome

Track type #1:

Points

Track geometry
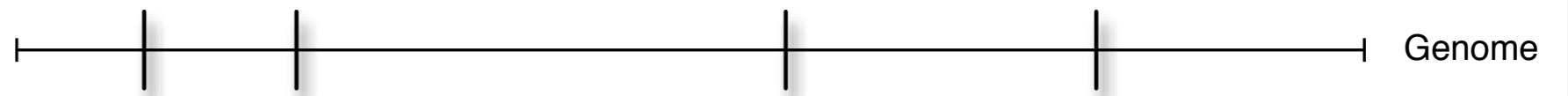
Track type #2:

Segments
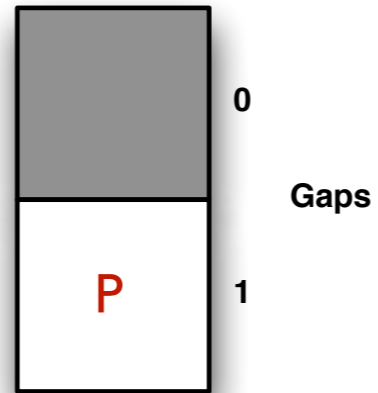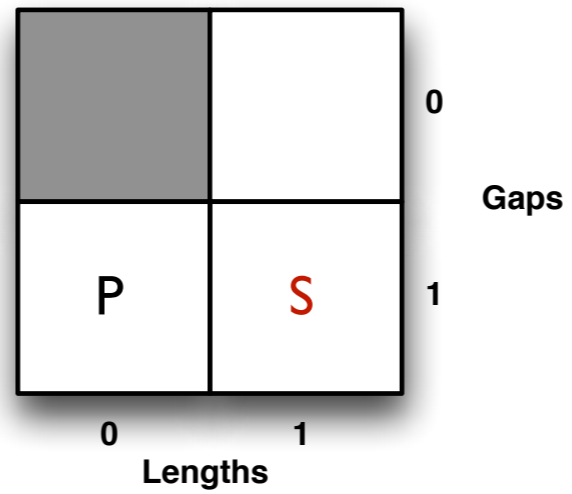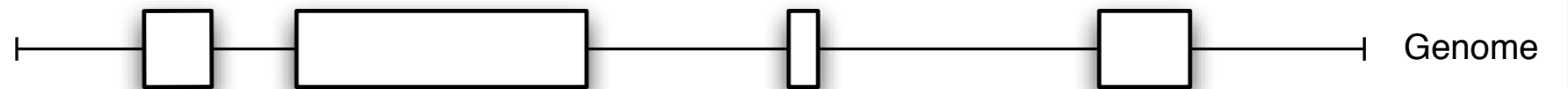

Track geometry

Track type #3:

Genome partition
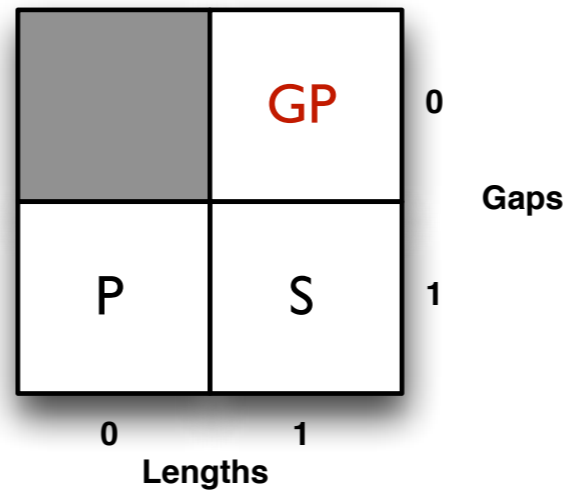
Track geometry

Track
type #4:

Valued
points
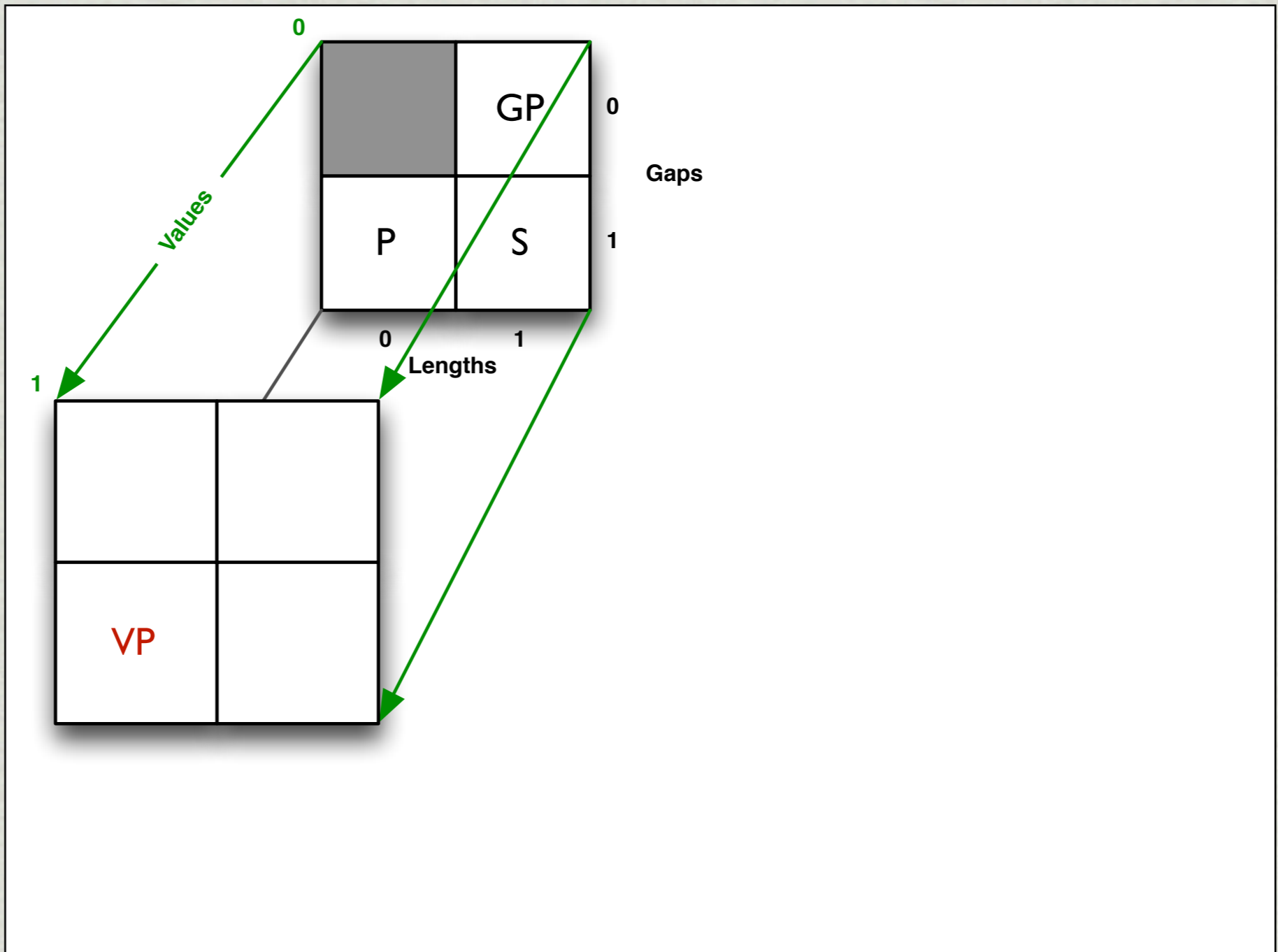
Track
geometry
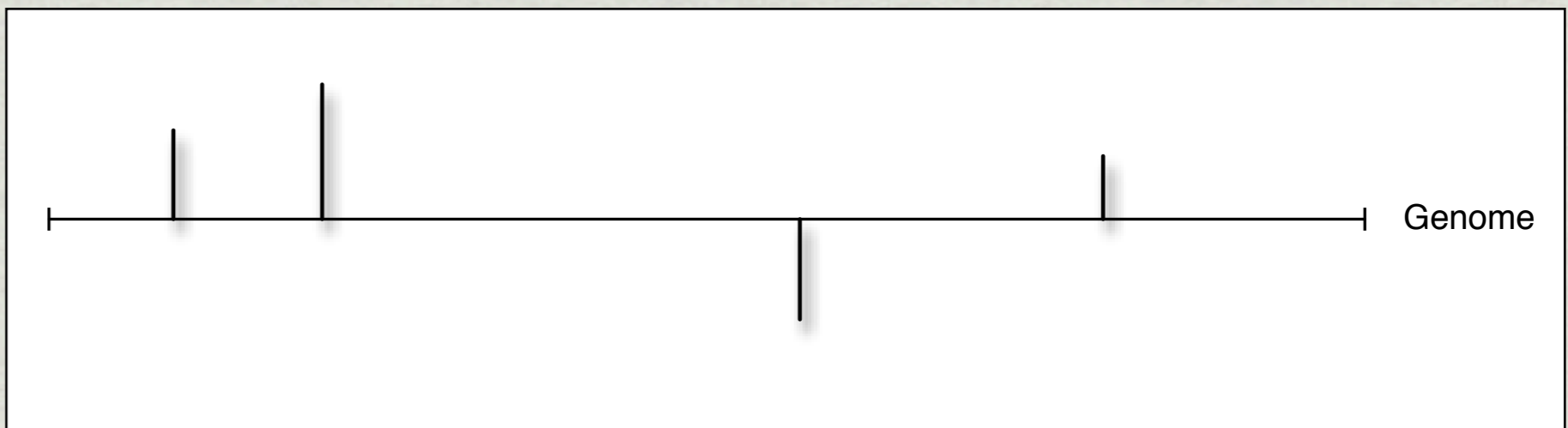
Track type #5:

Valued segments
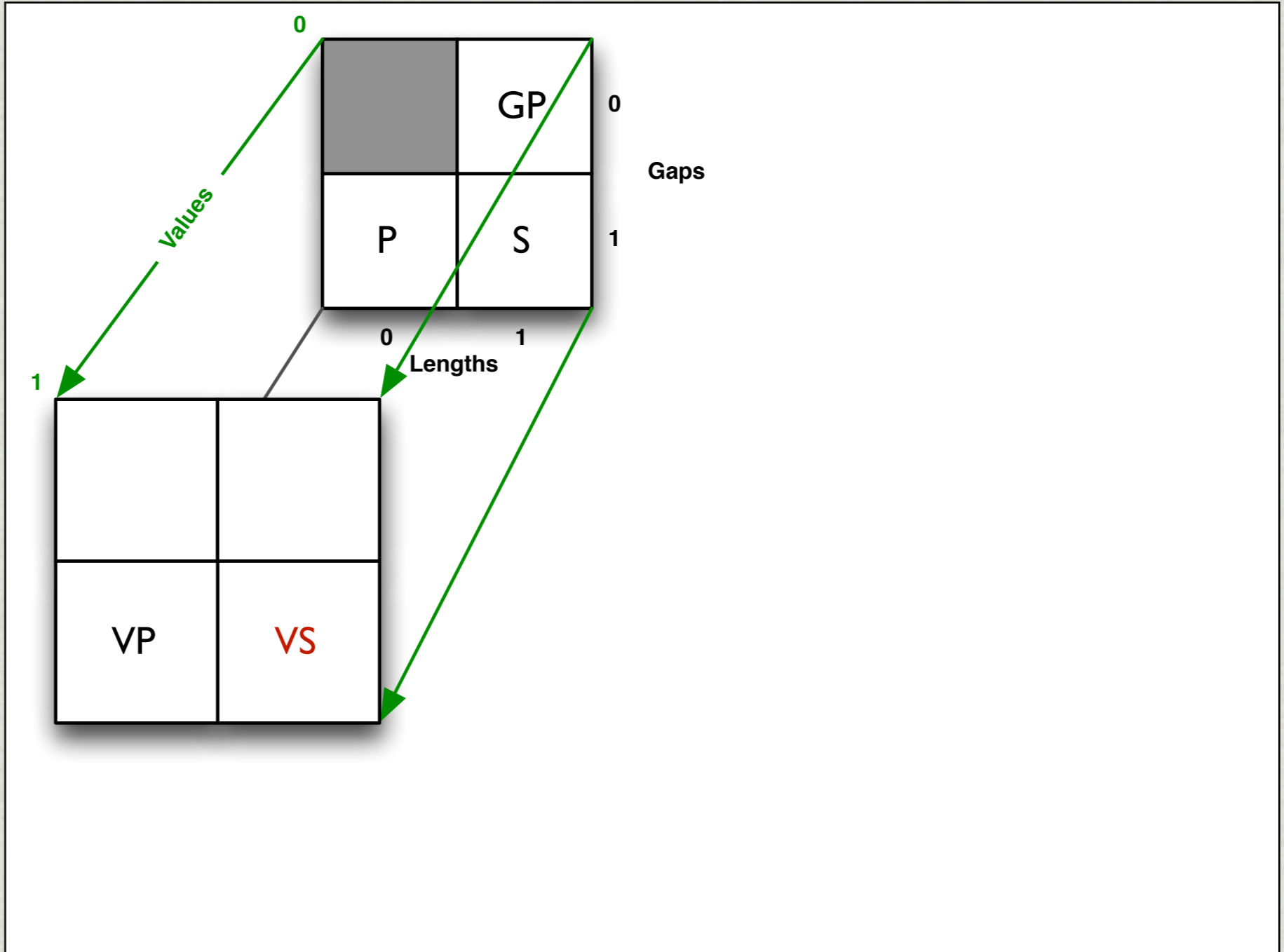
Track geometry
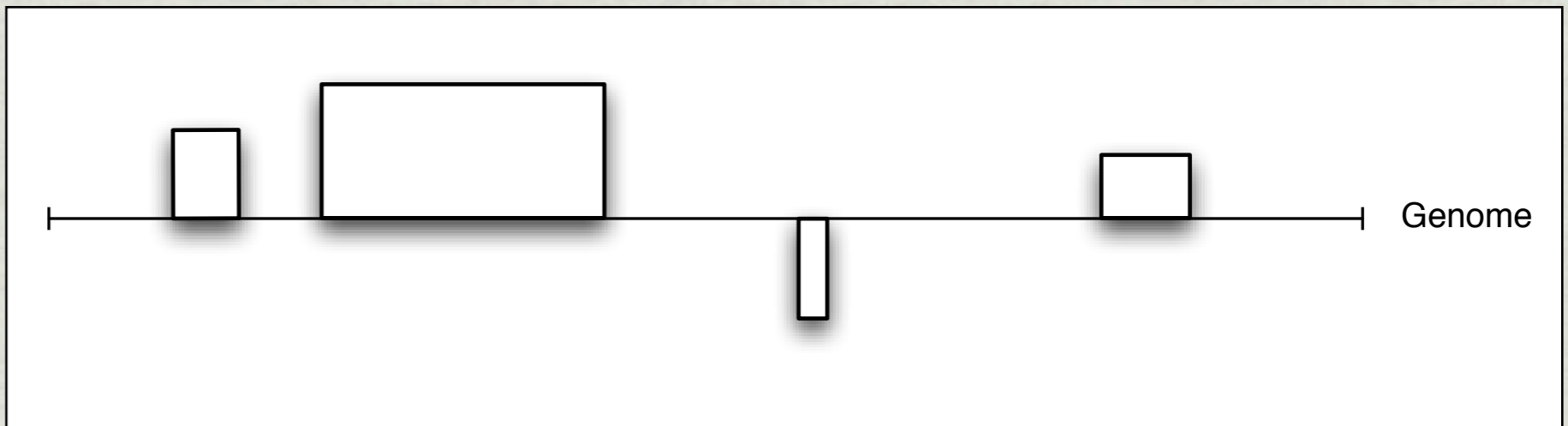
Track
type #6:

Step
function
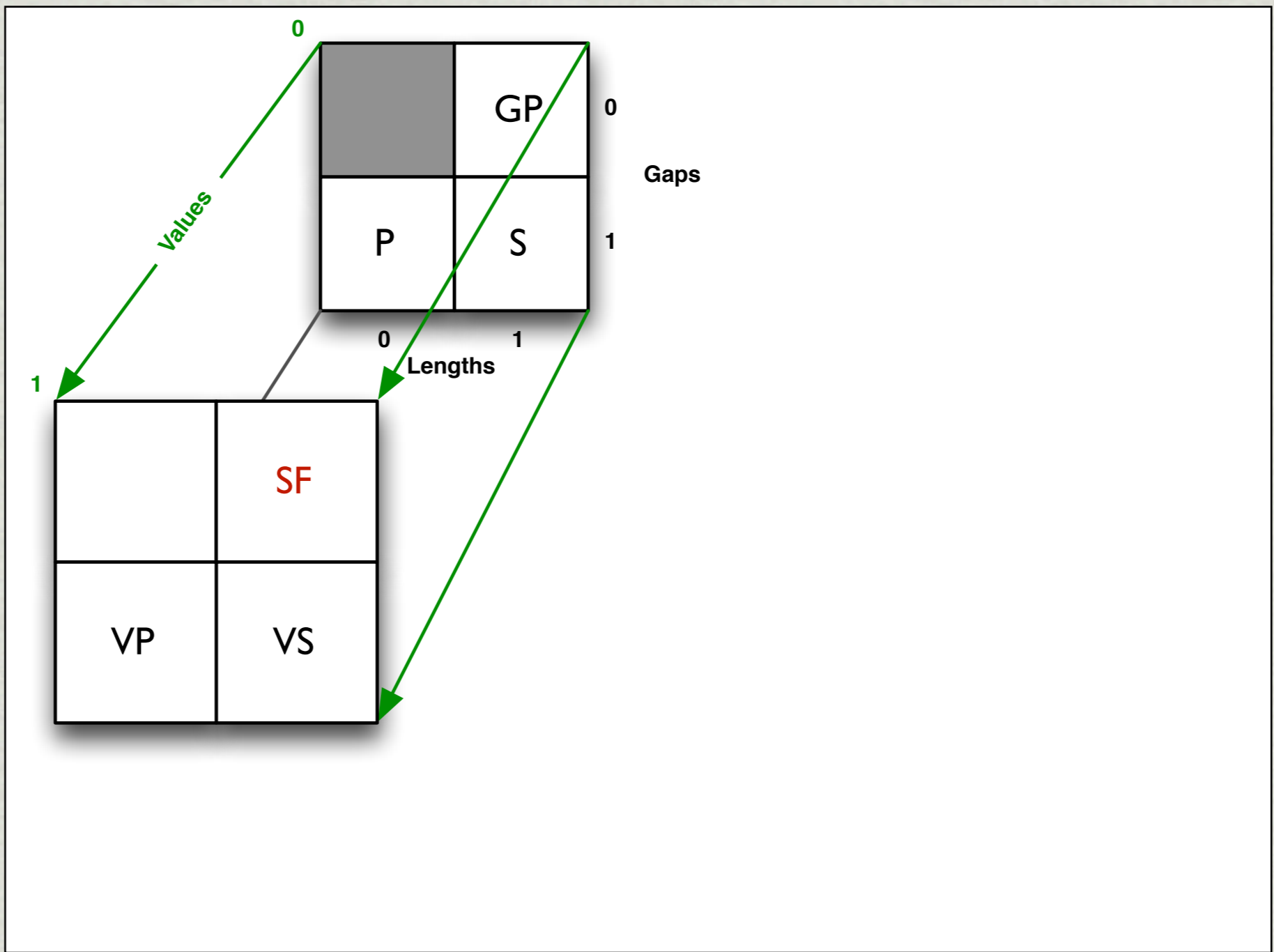
Track
geometry
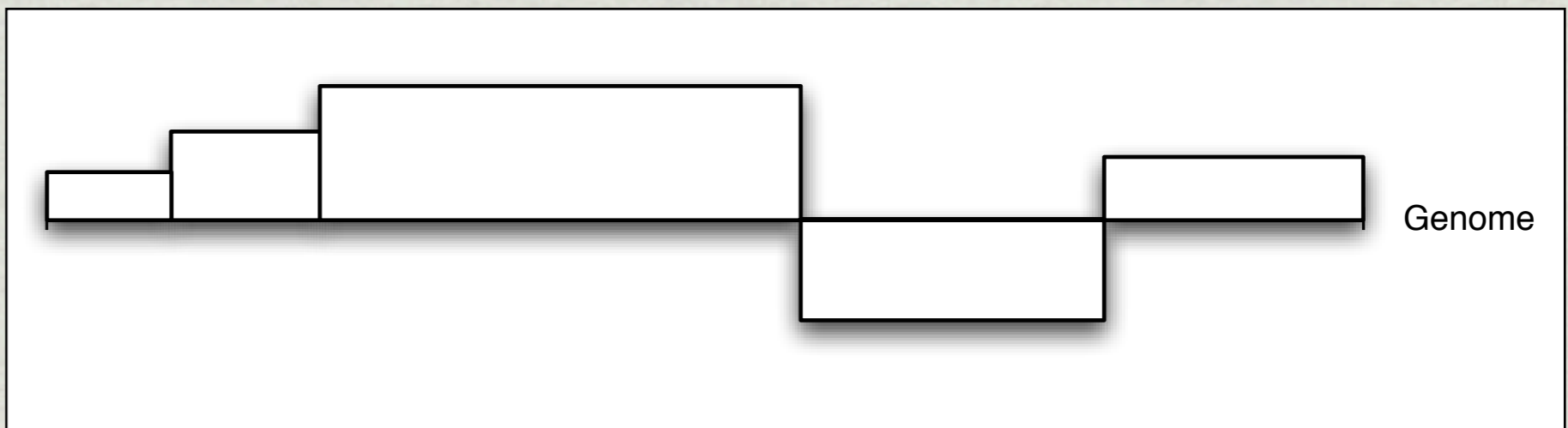
Track type #7:

Function
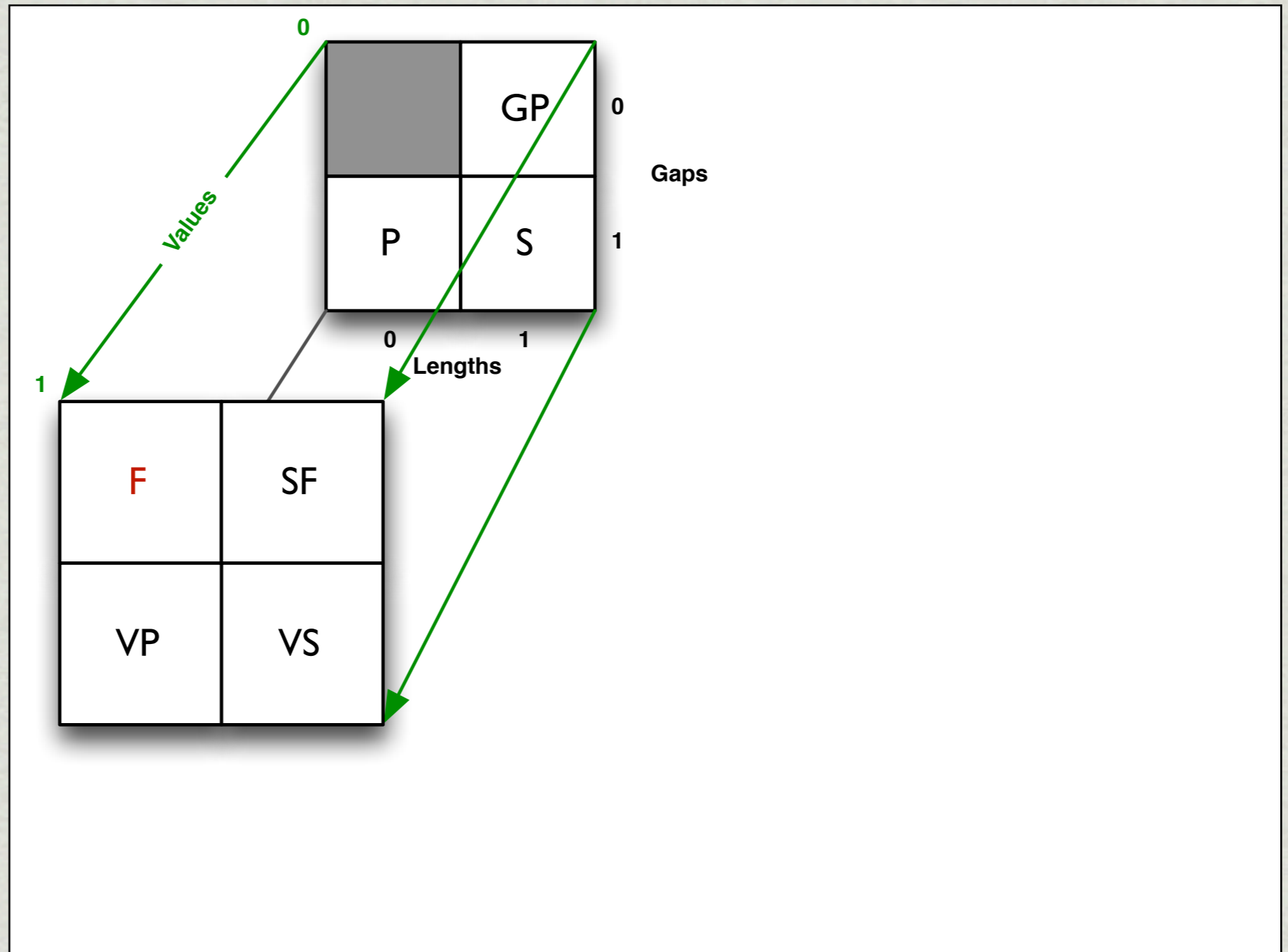
Track geometry
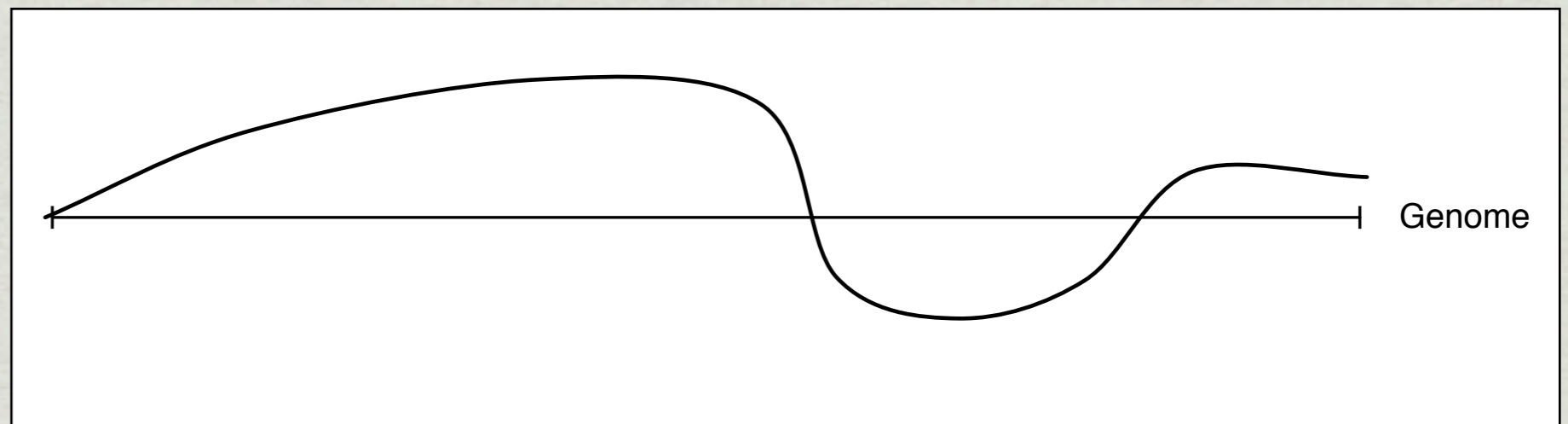
Track type #8-15:

Linked track types
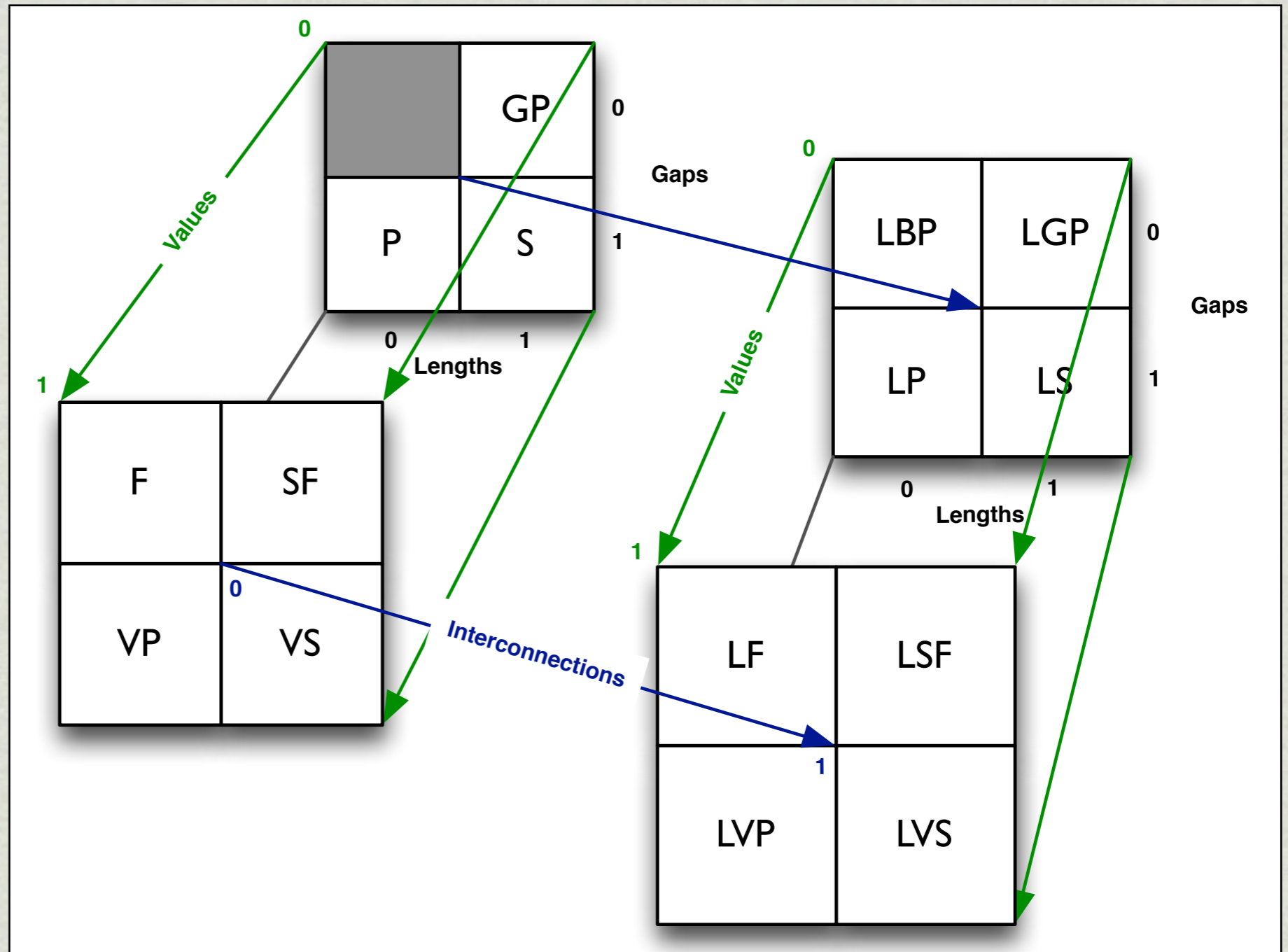
Track geometry
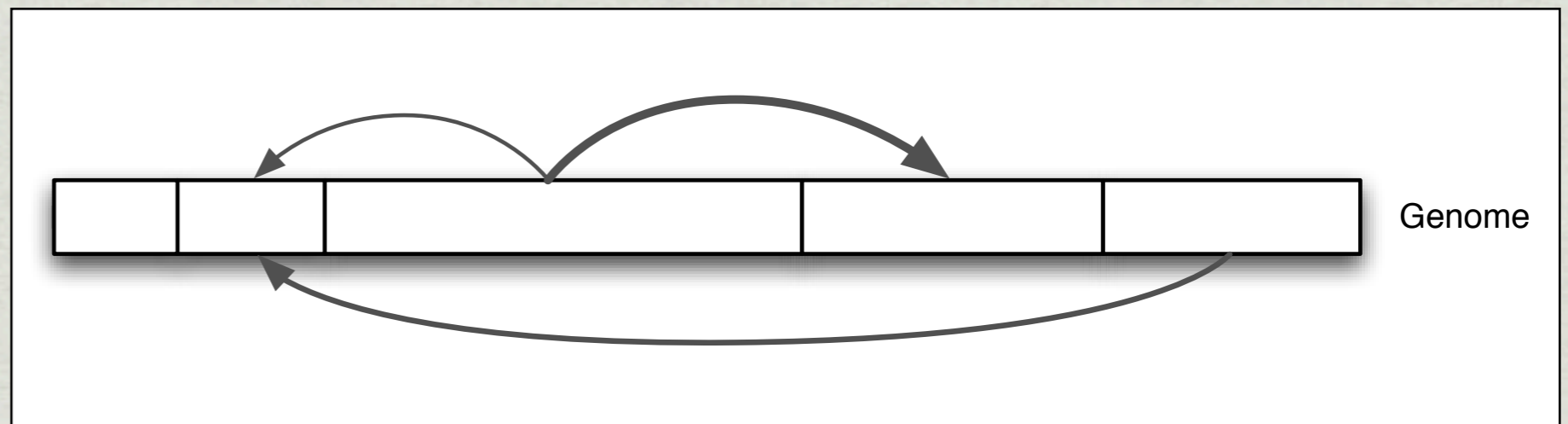
All 15 track types

Basic track types — Extended track types
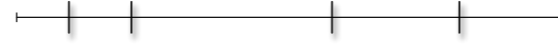
Points (P) — Linked Points (LP)
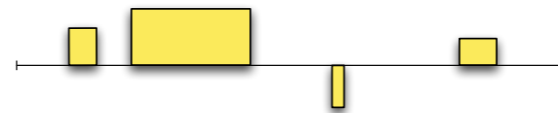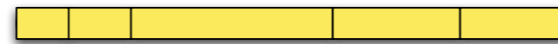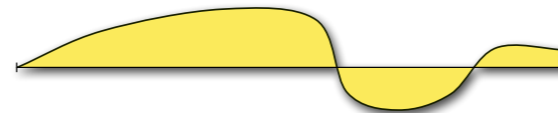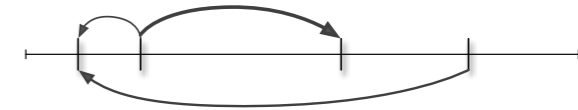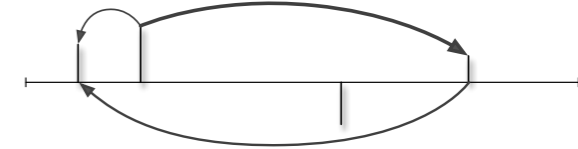
Valued Points (VP) — Linked Valued Points (LVP)
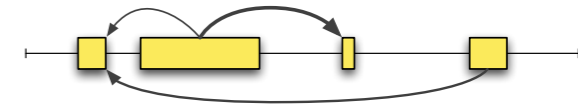
Segments (S) — Linked Segments (LS)
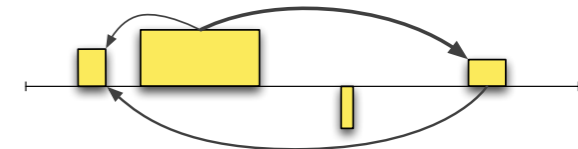
Valued Segments (VS) — Linked Valued Segments (LVS)
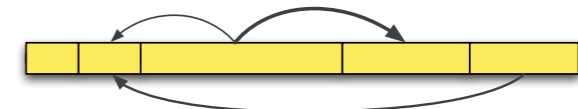
Genome Partition (GP) — Linked Genome Partition (LGP)
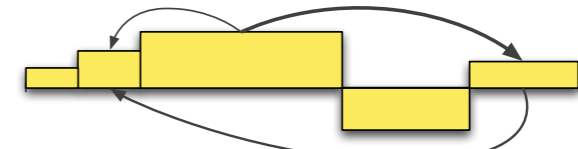
Step Function (SF) — Linked Step Function (LSF)
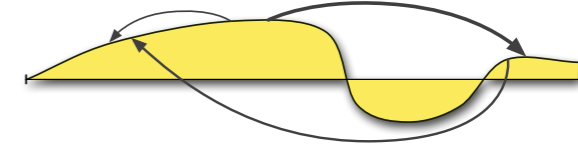
Function (F) — Linked Function (LF)
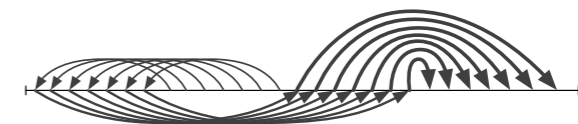
Linked Base Pairs (LBP)

Track types relate to analysis questions

P	P	Different frequencies?
P	P	Located nearby?
P	S	Located inside?
P	S	Located nonuniformly inside?
P	S	Located nearby?
S	S	Similar segments?
S	S	Overlap?
S	S	Located nearby?
F	F	Correlated?
P	F	Higher values at locations?
S	F	Higher values inside?
P	VS	Located in segments with high values?
S	VP	Higher values inside segments?
VP	VP	Nearby values similar?
P	VS (c/c)	Located in case segments
VS (c/c)	S	Preferential overlap?
VP (cat)	VS (cat)	Category pairs differentially co-located?
LGP	P	Colocalized in 3D?

Track types supported by different formats

Format	Ref.	Data	Repr.	P	S	VP	VS	GP	SF	F	L	Strand	#Cols	Value type
GFF3/GTF	[2]	General	Tab.	√(1)	√	√(1)	√				(2)	√	9	Float (3)
BED/bigBed	[4]	General	Tab./Bin.	√(1)	√	√(1)	√				(2)	√	3-12	Int(0-1000)/string (4)
BED15	[4]	Microarray	Tab.			√(1)	√				(2)	√	15	List of floats (5)
bedGraph	[4]	General	Tab.			√(1)	√						4	Float
WIG/bigWig (fixedStep)	[8]	General	Tab./Bin.			√	√		√	√			1	Float
WIG/bigWig (variableStep)	[8]	General	Tab./Bin.			√	√						2	Float
CNT	[36]	Copy number	Tab.			√							4	Float
Personal Genome SNP	[4]	Variation	Tab.			√(1)	√						7	String (6)
VCF	[37]	Variation	Tab.			√	√						≥ 8	String (6) (3)
GVF	[6]	General/Variation	Tab.	√(1)	√	√(1)	√				(2)	√	9	Float (3)
PSL	[4]	Alignment	Tab.		√		√					√	21	Int (7)
SAM/BAM	[38]	Alignment	Tab./Bin.		√		√					√	11	Int/string (8)
BioHDF	[39]	Alignment	Bin.		√		√					√	11	Int/string (8)
MAF	[4]	Multiple Alignment	Tab.		√		√				(9)	√	2-7	Float/string (8)
FASTA	[40]	Sequence	Text						√				N/A	Char
DAS XML	[12]	General	XML	√(1)	√	√(1)	√				(2)	√	N/A	Float
BioXSD 1.0	[16]	General	XML	√(10)	√(10)	√(10)	√(10)				√(11)	√	N/A	Float (12)
USeq	[19]	General	Bin.	√	√	√	√					√	N/A	Int/float/string
Genomedata	[41]	General	Bin.			√	√		√	√			N/A	Int/float/char

Why all the formats?

1. Different columns

2. Underlying structural differences

3. **Format-specific characteristics**

4. Tool-specific format requirements

5. Performance issues

Format-specific characteristics

- GFF format:
    - 1-based coordinates
    - Circular genomes
    - Format-specific headers

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- Galaxy tool (BedTools):

  - intersect multiple sorted BED files

- Segtools toolkit:

  - variant of BED where column 4 is interpreted as a category, rather than a name

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Performance issues

- Binary (indexed) versions of tabular formats are commonly created to improve performance:
  - SAM => BAM
  - BED => bigBed
  - WIG => bigWig
  - Any interval => Tabix

Introducing GTrack v1.0...

GTrack example (simple)

```
chr1        2396586      2827369
chr1        92014277     93306499
chr1        100983315    101455310
chr1        116832232    116909542
chr1        190733439    190814781
chr1        199128354    199336605
...
```

GTrack example (intermediate)

```
##gtrack version: 1.0
##track type: valued segments
##value type: binary
##sorted elements: true
##1-indexed: true
##end inclusive: true
##my custom header: Yes!
###seqid     start         end           value
chr1         2396587       2827369       1
chr1         92014278      93306499      1
chr1         100983316     101455310     0
chr1         116832233     116909542     0
chr1         190733440     190814781     1
chr1         199128355     199336605     0
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chr1        100983316   101455310    0
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chr1        190733440   190814781    1
chr1        199128355   199336605    0
...
```

Flexible columns

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chr1          2396587      2827369      1
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chr1          100983316    101455310    0
chr1          116832233    116909542    0
chr1          190733440    190814781    1
chr1          199128355    199336605    0
...
```

Supports all 15
track types

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chr1        190733440   190814781   1
chr1        199128355   199336605   0
...
```

Format-specific characteristics, custom headers

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chr1         92014278       93306499        1
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chr1         116832233      116909542       0
chr1         190733440      190814781       1
chr1         199128355      199336605       0
...
```

Tool-specific
format requirements

GTrack example (intermediate)

Specification of GTrack subtypes via URL
with automatic validation

```
##subtype url: http://my.edu/frmt.gtrack
chr1      2396587      2827369      1
chr1      92014278     93306499     1
chr1      100983316    101455310    0
chr1      116832233    116909542    0
chr1      190733440    190814781    1
chr1      199128355    199336605    0
...
```

GTrack example (advanced)

```
#Example track
##gtrack version: 1.0
##track type: linked genome partition
##edge weights: true
###end        id        edges
####seqid=chr1;start=1000;end=2000
1015          A         C=1.3,D=0.1
1060          B         A=1.0
1154          C         A=1.3
1267          D         .

...
```

GTrack example (advanced)

```
#Example track
##gtrack version: 1.0
##track type: linked genome partition
##edge weights: true
###end    id        edges
####seqid=chr1;start=1000;end=2000
1015      A         C=1.3,D=0.1
1060      B         A=1.0
1154      C         A=1.3
1267      D         .

...
```

Specification of edges with weights

GTrack example (advanced)

```
#Example track
##gtrack version: 1.0
##track type: linked genome partition
##edge weights: true
###end       id       edges
####seqid=chr1;start=1000;end=2000
1015         A         C=1.3,D=0.1
1060         B         A=1.0
1154         C         A=1.3
1267         D         .

...
```

Specification of bounding regions (to exclude missing data / assembly gaps)

GTrack performance

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- Converts to/from many common formats

- Planned to move to PyTables:

  - HDF5-based, compressed storage

  - advanced indexing and querying

  - solid performance

  - several tracks in one file

Galaxy relevance of GTrack

- 15 track types (including 3D type data)!

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- Everything specified in the file itself, interoperable with other systems

- GTrack fully integrated with the HyperBrowser, along with a set of GTrack-specific tools

Galaxy relevance of GTrackCore

- HDF5-based binary file as history element

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- Fast performance (after indexing)

Galaxy relevance of GTrackCore

- HDF5-based binary file as history element

- Fast performance (after indexing)

- Library of operations, analysis, statistics to ease tool development

Interested?

We need more developers!

from http://xkcd.com

Nothing ventured, nothing gained...

Acknowledgements

More info

Gundersen S, Kalas M, Abul O, Frigessi A, Hovig E, Sandve GK: *Identifying elemental genomic track types and representing them uniformly.* BMC Bioinformatics 2011, 12:494.

Specification available at:
www.gtrack.no

GTrackCore source code:
https://github.com/i000/gtrackcore