

SigProfilerAssignment

Presented by:

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Cancer Genome Analysis

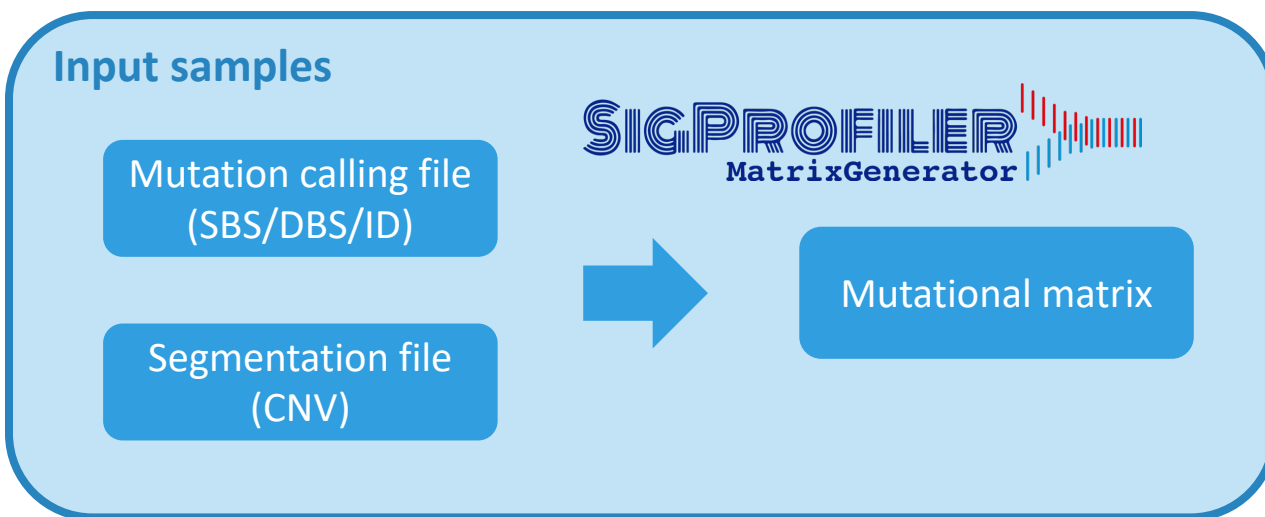
12 – 16 September 2022 – Virtual course



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Overview of SigProfilerAssignment



Overview of SigProfilerAssignment



Input samples

Mutation calling file
(SBS/DBS/ID)

Segmentation file
(CNV)



Mutational matrix

$$\begin{matrix} \mathbf{M} \\ t \times n \end{matrix} = \begin{matrix} \mathbf{S} \\ t \times k \end{matrix} \times \begin{matrix} \mathbf{A} \\ k \times n \end{matrix}$$

Reference mutational signatures

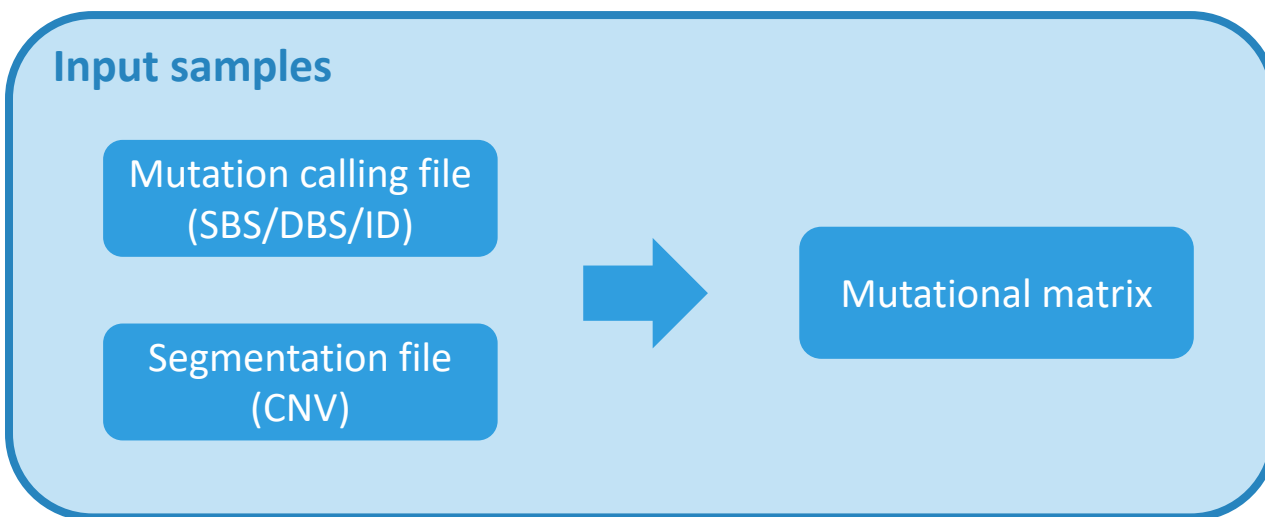


v1, v2, v3, v3.1, v3.2, v3.3

GRCh37/38, mm9/10, rn6

Genome / Exome

Overview of SigProfilerAssignment



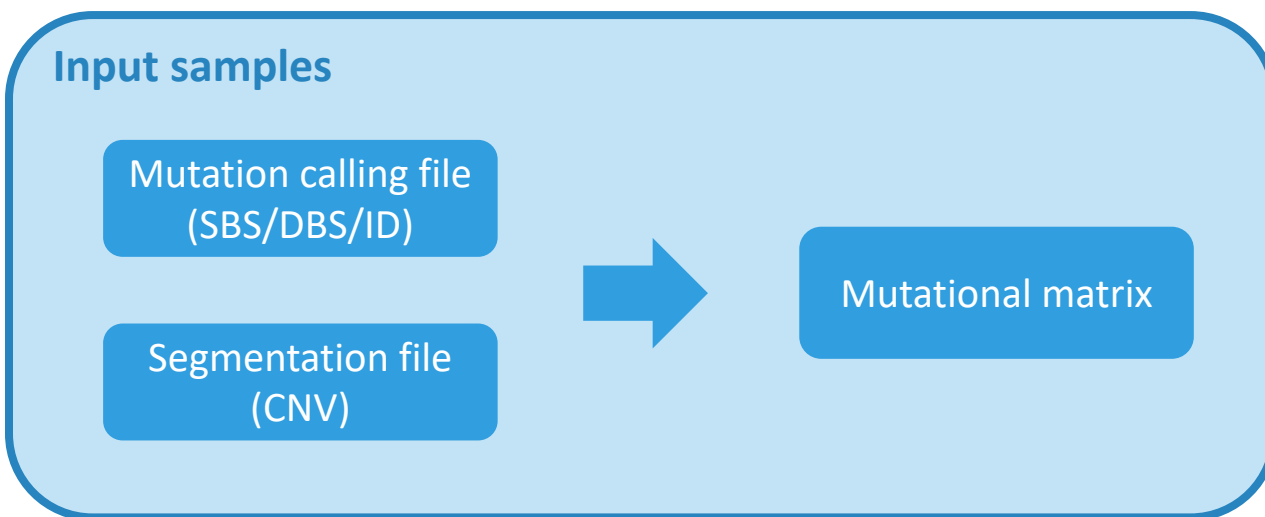
$$\mathbf{M} = \mathbf{S} \times \mathbf{A}$$

$t \times n$ $t \times k$ $k \times n$

t mutational contexts
 n samples
 k signatures



Overview of SigProfilerAssignment

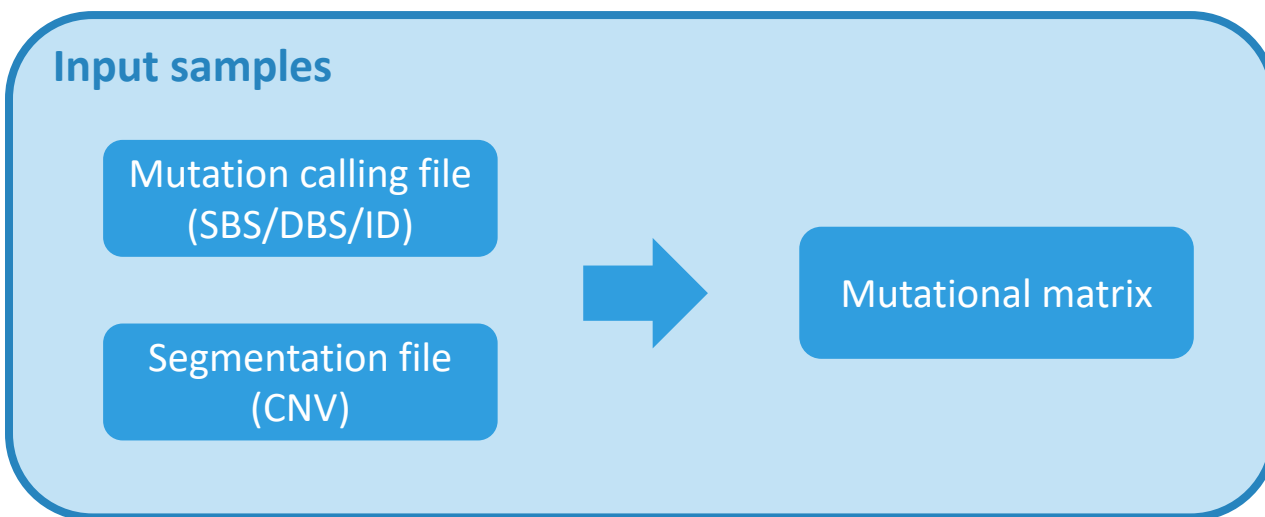


$$\underset{t \times n}{\mathbf{M}} = \underset{t \times k}{\mathbf{S}} \times \underset{k \times n}{\mathbf{A}}$$

t mutational contexts
 n samples
 k signatures



Overview of SigProfilerAssignment



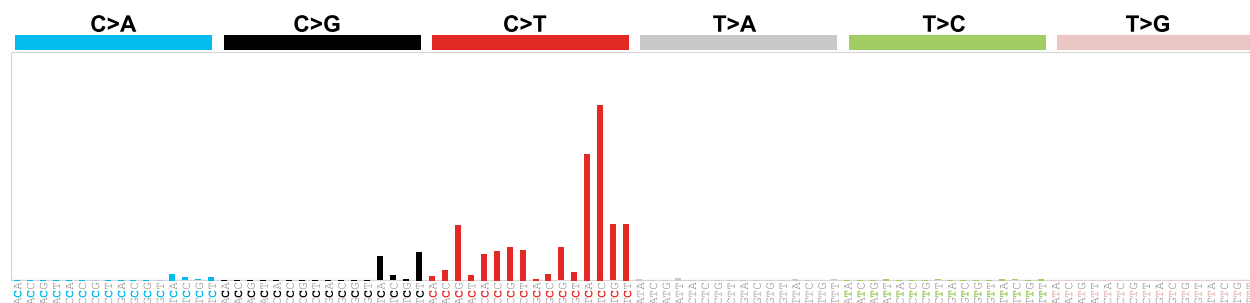
$$\underset{t \times n}{\mathbf{M}} = \underset{t \times k}{\mathbf{S}} \times \boxed{\underset{k \times n}{\mathbf{A}}}$$

t mutational contexts
 n samples
 k signatures



Assigning reference signatures to individual samples

M



Mutational profile

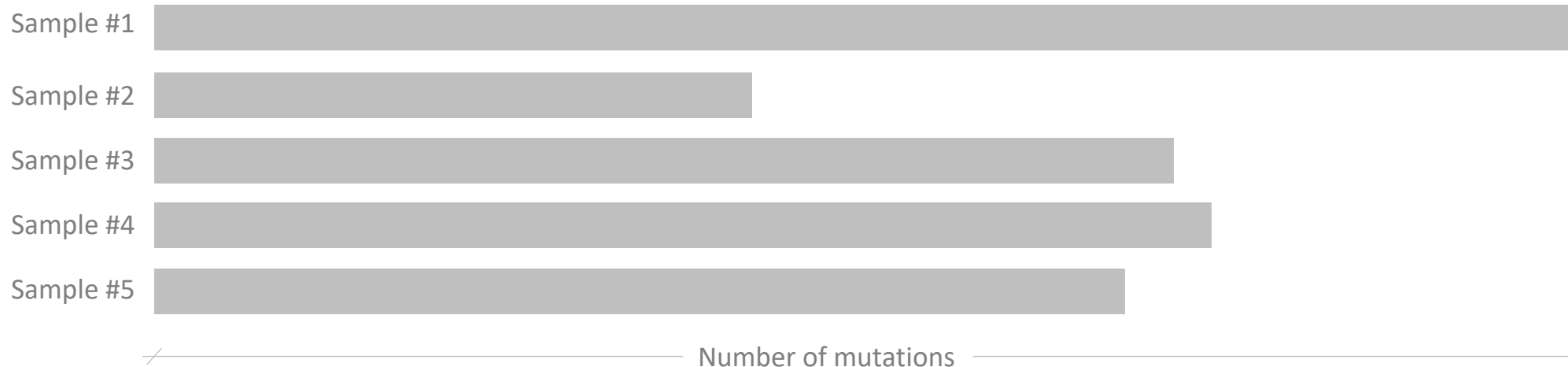
#CHROM	POS	FILTER	REF	ALT
1	809687	PASS	G	C
1	819245	PASS	G	T
1	1911011	PASS	C	G
1	2112413	PASS	T	C
1	2927666	PASS	A	G
1	3359791	PASS	C	T
1	4347912	PASS	G	A
1	4961889	PASS	G	C
1	5949138	PASS	C	T
1	7806339	PASS	A	C
1	9648435	PASS	G	A
1	9705025	PASS	C	T

Mutation calling file

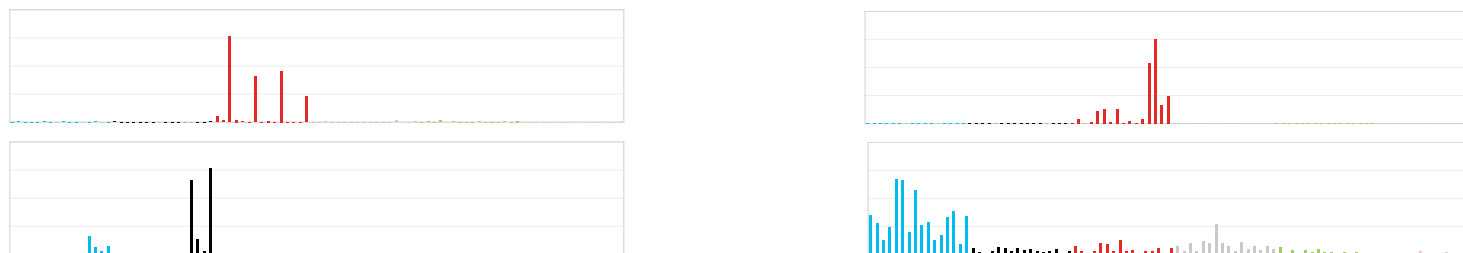


Assigning reference signatures to individual samples

M



S

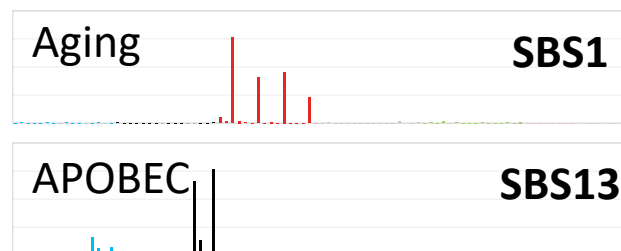


Assigning reference signatures to individual samples

M



S

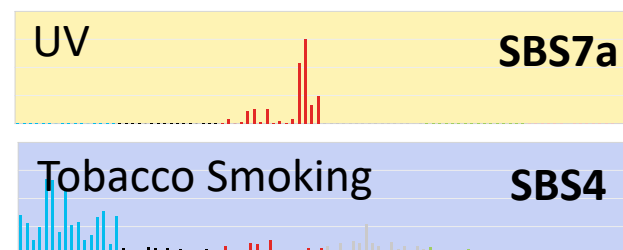
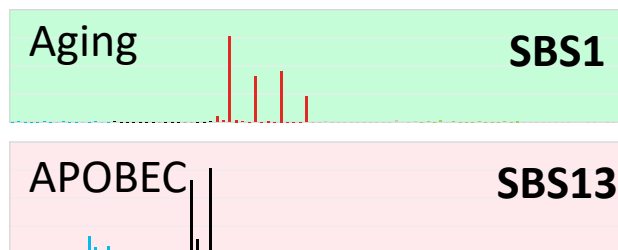


Assigning reference signatures to individual samples

M

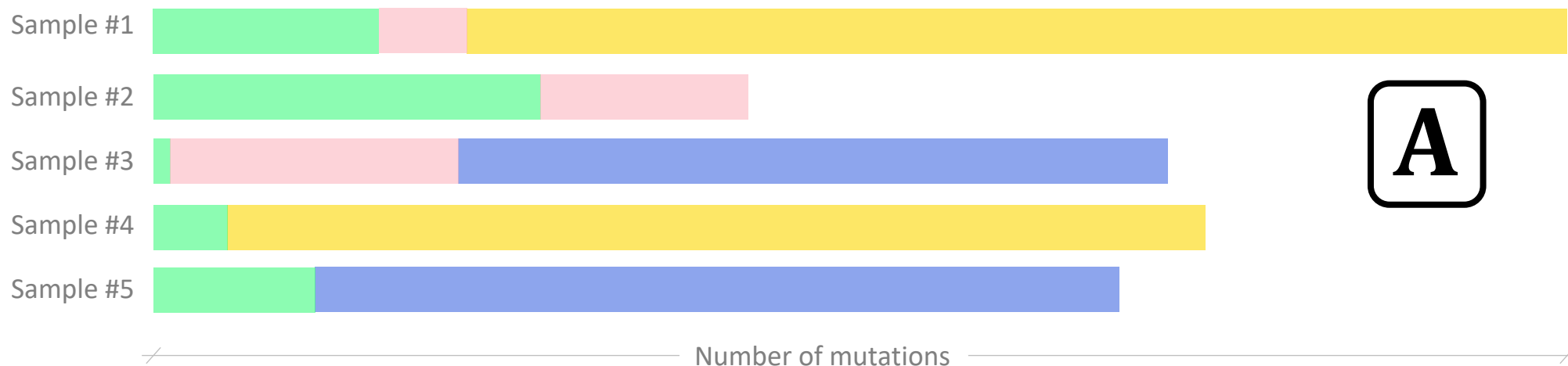


S

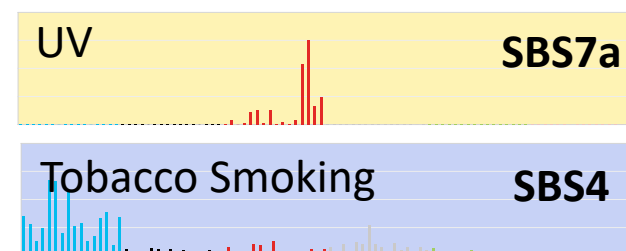
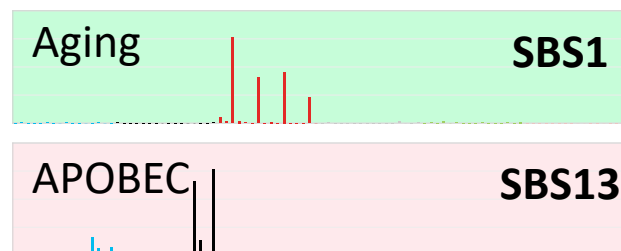


Assigning reference signatures to individual samples

M



S



Useful links – SigProfilerAssignment

- GitHub repository
<https://github.com/AlexandrovLab/SigProfilerAssignment>
- Python package (PyPI)
<https://pypi.org/project/SigProfilerAssignment/>
- COSMIC Mutational Signatures website
<https://cancer.sanger.ac.uk/signatures/>