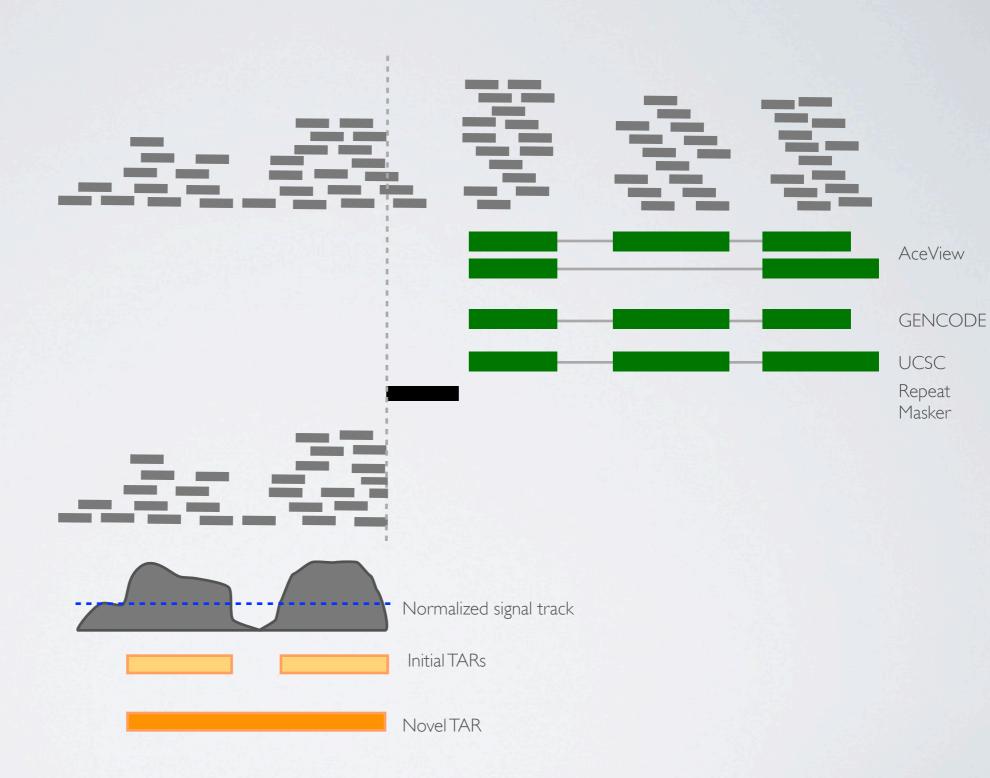
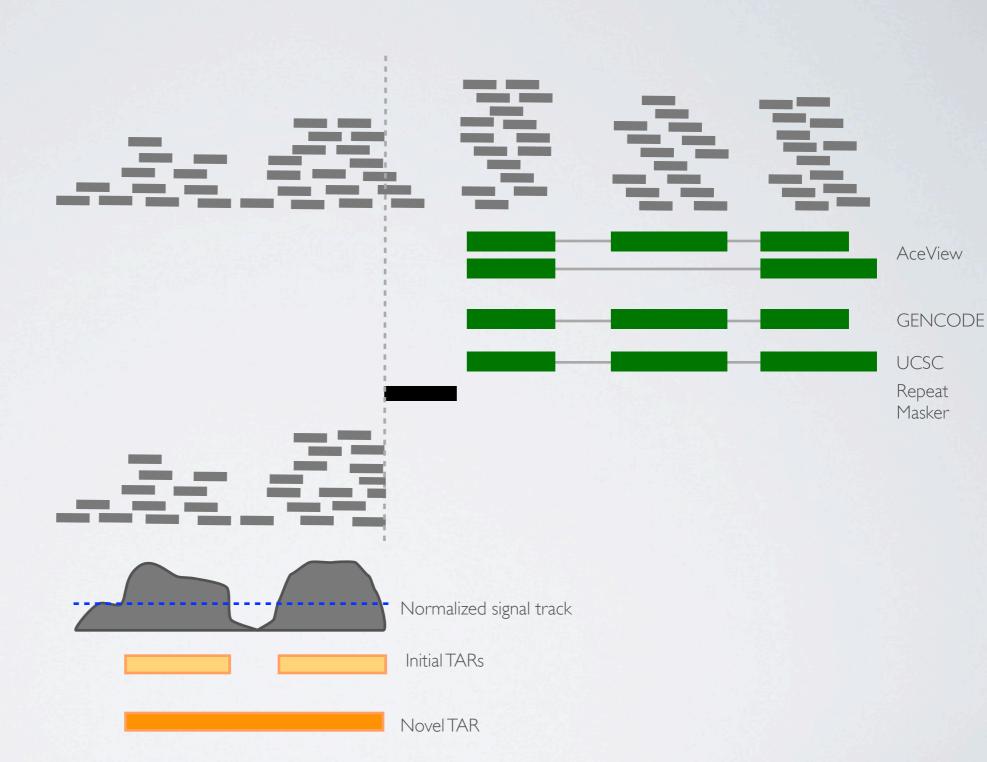
NOVEL TRANSCRIPTIONALLY ACTIVE REGIONS (TARS)

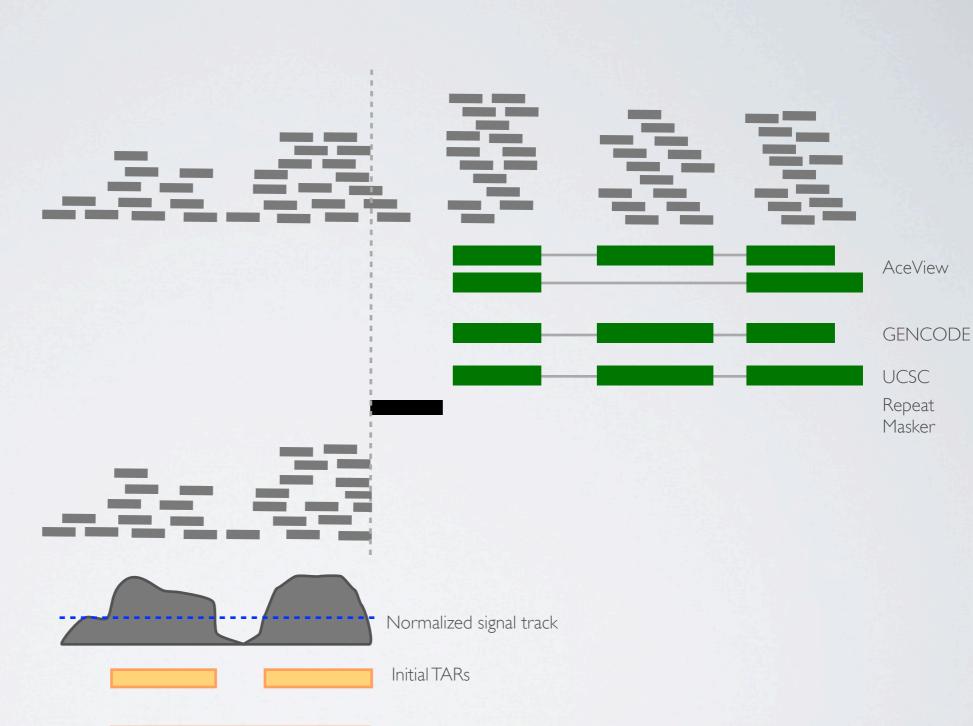
Andrea Sboner - 2011.04.08



 Reads overlapping gene annotation set --AceView, GENCODE, UCSC, and repetitive regions are excluded

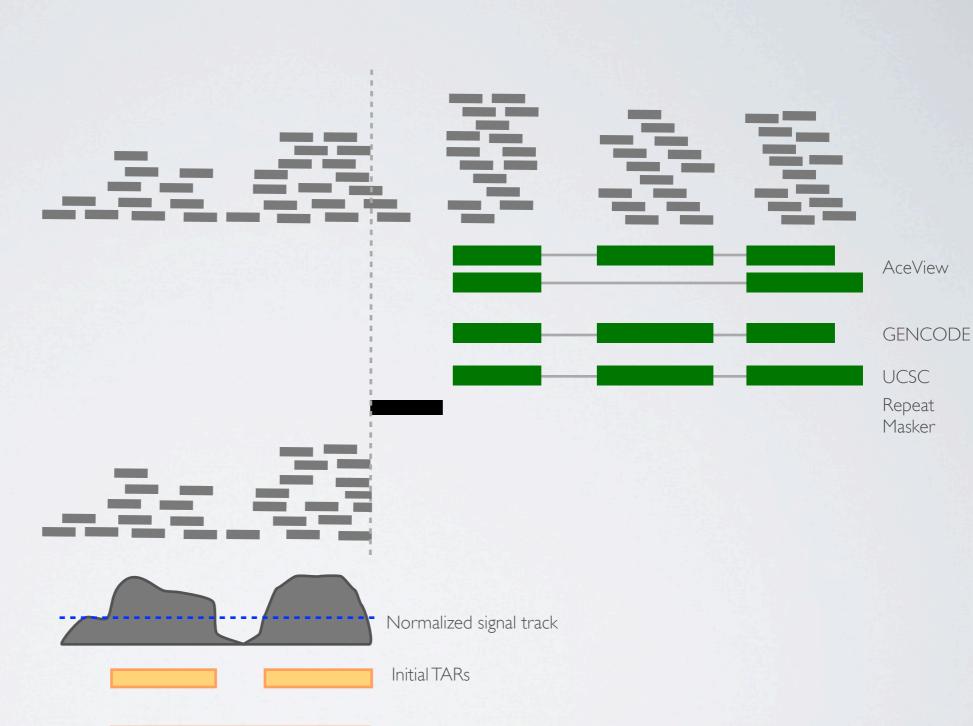


- Reads overlapping gene annotation set --AceView, GENCODE, UCSC, and repetitive regions are excluded
- Reads from all samples pooled together



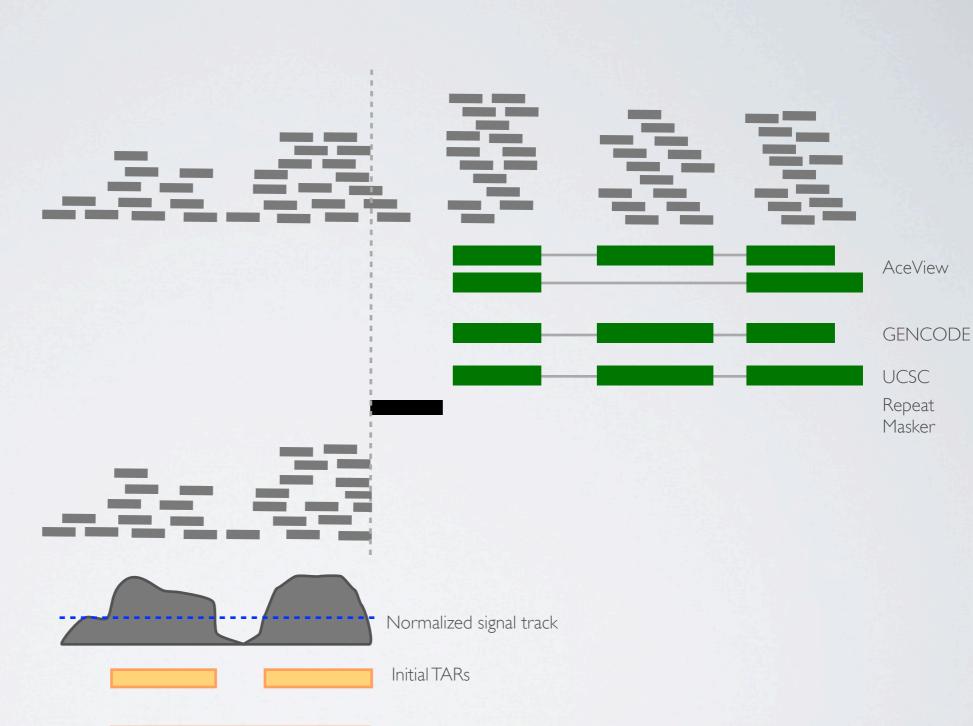
Novel TAR

- Reads overlapping gene annotation set --AceView, GENCODE, UCSC, and repetitive regions are excluded
- Reads from all samples pooled together



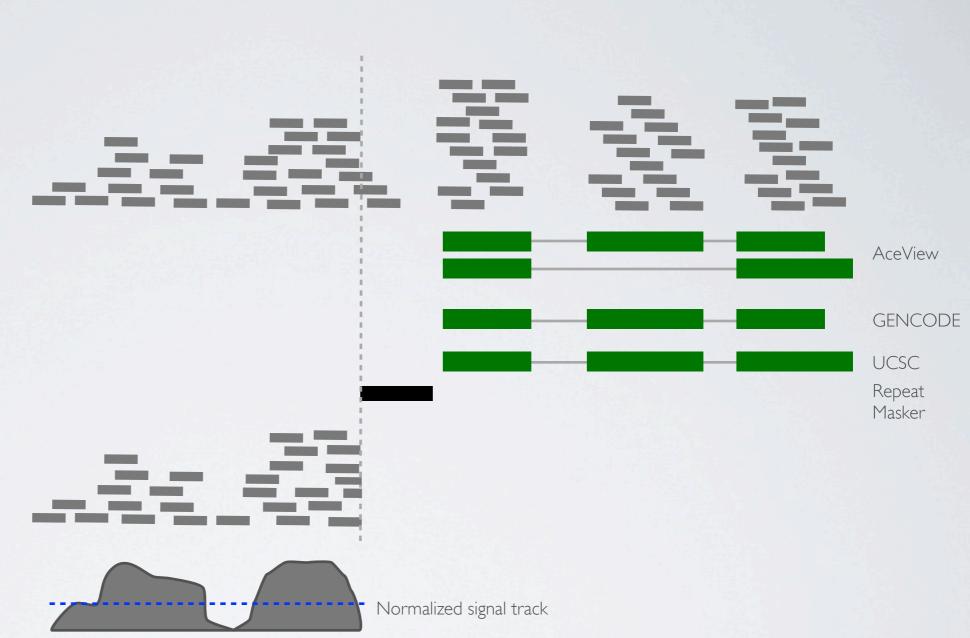
Novel TAR

- Reads overlapping gene annotation set --AceView, GENCODE, UCSC, and repetitive regions are excluded
- Reads from all samples pooled together



Novel TAR

- Reads overlapping gene annotation set --AceView, GENCODE, UCSC, and repetitive regions are excluded
- Reads from all samples pooled together



Initial TARs

Novel TAR

Max-gap, min-run algorithm to identify TARs

$$|\vec{L}| = 0.129Kb$$

$$r(g) = \frac{n(g)}{l(g)*M}$$

$$|\vec{L}| = 0.129Kb$$

$$r(g) = \frac{n(g)}{l(g)*M} \qquad x(g) = log_2(r(g)+1)$$

$$|\vec{L}| = 0.129Kb$$

$$r(g) = \frac{n(g)}{l(g)*M} \qquad x(g) = log_2(r(g)+1)$$

• From exon expression values: Determine the median value of each gene:

$$\tilde{x}(g)$$
 $x_c = quantile(\vec{x}, 0.05)$
 $\forall \tilde{x}(g) > 0$
 $r_c = 2^{x_c} - 1$
 $|\vec{L}| = 0.129Kb$
 $t_{bqr} = |\vec{L}| \cdot r_c$

$$r(g) = \frac{n(g)}{l(g)*M} \qquad x(g) = log_2(r(g)+1)$$

- From exon expression values: Determine the median value of each gene:
 - Computed the 5th percentile of expressed genes:

$$ilde{x}(g)$$

$$x_c = quantile(\vec{\tilde{x}}, 0.05)$$
$$\forall \tilde{x}(g) > 0$$

$$r_c = 2^{x_c} - 1$$

$$|\vec{L}| = 0.129Kb$$

$$t_{bgr} = |\vec{L}| \cdot r_c$$

$$r(g) = \frac{n(g)}{l(g)*M} \qquad x(g) = log_2(r(g)+1)$$

- From exon expression values:

 Determine the median value of each gene:
 - Computed the 5th percentile of expressed genes:
 - Compute the corresponding RPKM value:

$$\tilde{x}(g)$$

$$x_c = quantile(\vec{\tilde{x}}, 0.05)$$
$$\forall \tilde{x}(g) > 0$$

$$r_c = 2^{x_c} - 1$$

$$|\vec{L}| = 0.129Kb$$

$$t_{bgr} = |\vec{L}| \cdot r_c$$

$$r(g) = \frac{n(g)}{l(g)*M} \qquad x(g) = log_2(r(g)+1)$$

• From exon expression values: Determine the median value of each gene:

$$\tilde{x}(g)$$

- Computed the 5th percentile of expressed genes:
- Compute the corresponding RPKM value:
- Consider the median length of GENCODE exons:

$$x_c = quantile(\vec{x}, 0.05)$$

$$\forall \vec{x}(g) > 0$$

$$r_c = 2^{x_c} - 1$$

$$|\vec{L}| = 0.129Kb$$

$$t_{bgr} = |\vec{L}| \cdot r_c$$

$$r(g) = \frac{n(g)}{l(g)*M} \qquad x(g) = log_2(r(g)+1)$$

• From exon expression values:

Determine the median value of each gene:

$$\tilde{x}(g)$$

Computed the 5th percentile of expressed genes:

$$x_c = quantile(\vec{\tilde{x}}, 0.05)$$
$$\forall \tilde{x}(g) > 0$$

Compute the corresponding RPKM value:

$$r_c = 2^{x_c} - 1$$

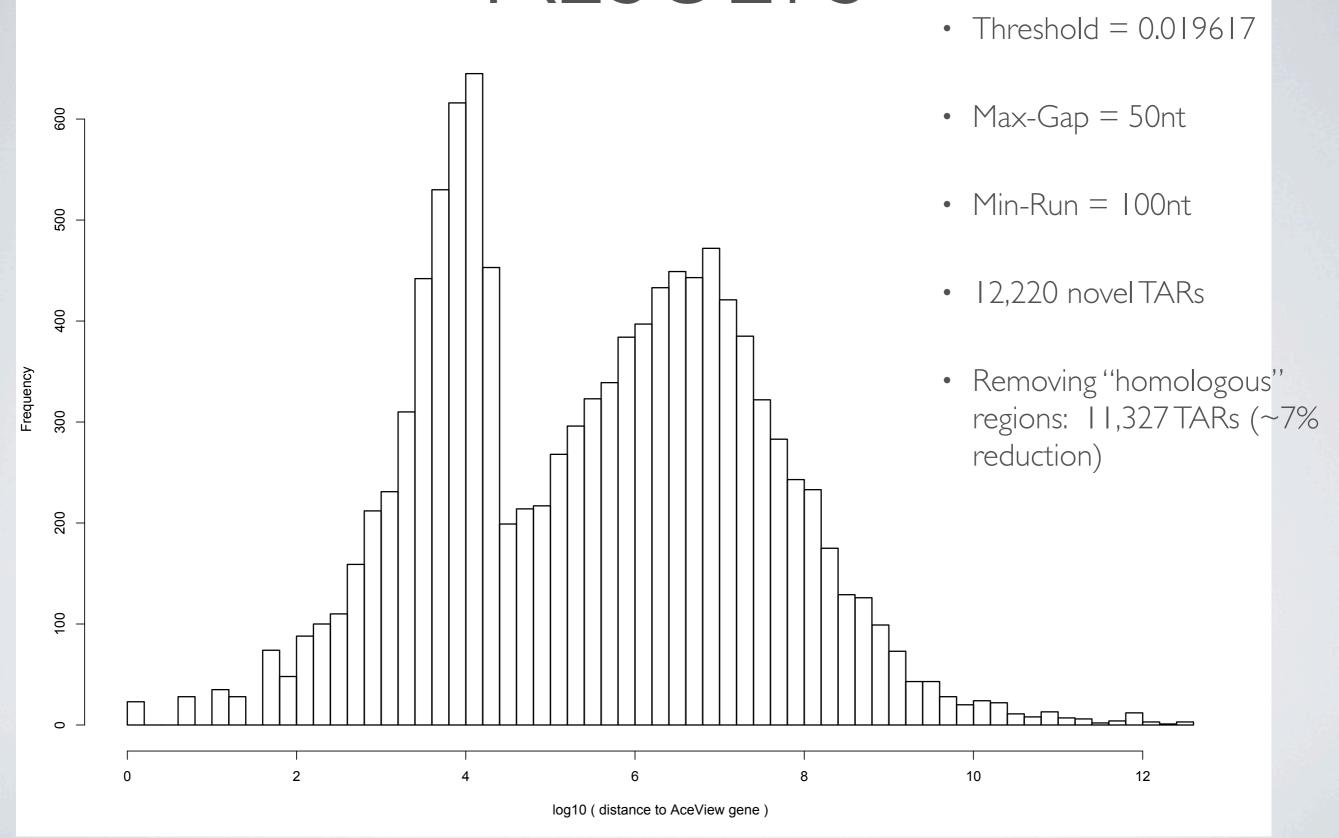
Consider the median length of GENCODE exons:

$$|\vec{L}| = 0.129Kb$$

• Define threshold (normalized per million mapped reads):

$$t_{bgr} = |\vec{L}| \cdot r_c$$

RESULTS



EXAMPLE

| | | p15.2 p14 | p13 p12.33 p | 12.2 p12.1 p1 | 1.22 p11.1 | q11.21 | q11.23 q21.1 | q21.2 | q21.3 q2 | 2.1 q22.2 | q22.3 q23. | .1 q23.2 c | 123.32 q24.1 q24 | 31 q25.1 | q25.2 q25.3 | q26.12 |
|--------------------------|--|---|--------------|------------------------|-----------------|---------------|------------------------|----------------|-----------------|------------|-----------------|---------------|--------------------|-------------|------------------|-----------|
| | Table Contra | 155 kb 3,260 kb 3,260 kb 3,260 kb 3,360 kb 3,360 kb | | | | | | | | | | | 3,350 Na | | | |
| | DATATHE DATATHE TRACK_ID ID Sample region region | 7.17 | | 7,70 | | 7717 | | 1 | | | 1 22 | | 7,77 | | 1,000 | |
| VFC | | E - | | -51 | 7 | | - : <u>-</u> | 1 | | Ħ | :- | 1 | - 1 | | | |
| V1C | | | | 1 | | - : | - 1- <u>- E</u> | - | | - | | | . <u> </u> | | | |
| STR | | No expre | ession | | | | | | | | | | | | | |
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| \$1C | | | | . #- | | 4 | 4 | Į : | 1 | H | | | Ę | | | |
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| MD | | Low expr | ression - | | | | =- | - | | | | | | | | |
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| IPC | | | | <u> </u> | | 1 | <u> </u> | 4 : | = = = | -17 | | | | | | |
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| CBC | | No expre | ession | | | | | | | | | | | | | |
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| RefSeq Genes | | | | | | | | | | | | | | | | |
| all.NOVEL.novel.0.019617 | | | BETO | 19 chr10 . 3280952 328 | 105 | | BED_21 chv10 .[3312391 | 3313912 pen | 241chr101122204 | 2513330547 | | | | | | |
| UCSC Genes | | | 560. | | | | | | | | | | I | · · · · · · | · · · · · | · · · · · |
| repeatMasker_hg19 | | (Nar-Tigger_3 Alab_SINE_Ala_3 | | WIRD_SINE_MIR_3 | MIRC_SINE_MIR_3 | Aur_SINE_Au_3 | 13 INTI IN 3 | INE_L2_3 L26_L | NELZ3 MIRS | INE HIR 3 | TR16E1_LTR_ERVL | 3 L24 LINE L3 | 2.3 MLT2A2_LTR_ERV | | -3 L1M83_UNE_L1_ | |