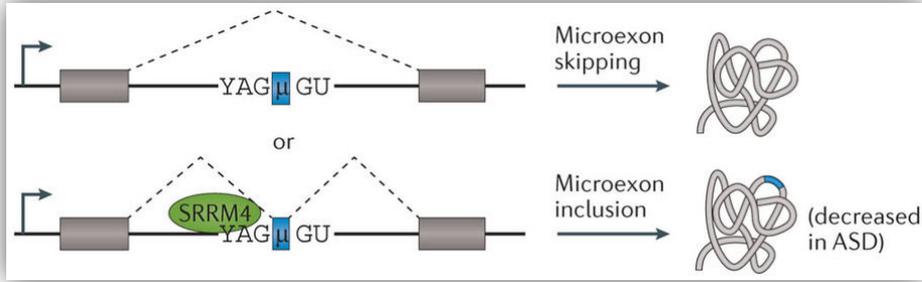


DISCERNs

DISCcovery of Exons from RNA-seq
Splice junction reads

Alternative splicing misregulation in neurological disorders

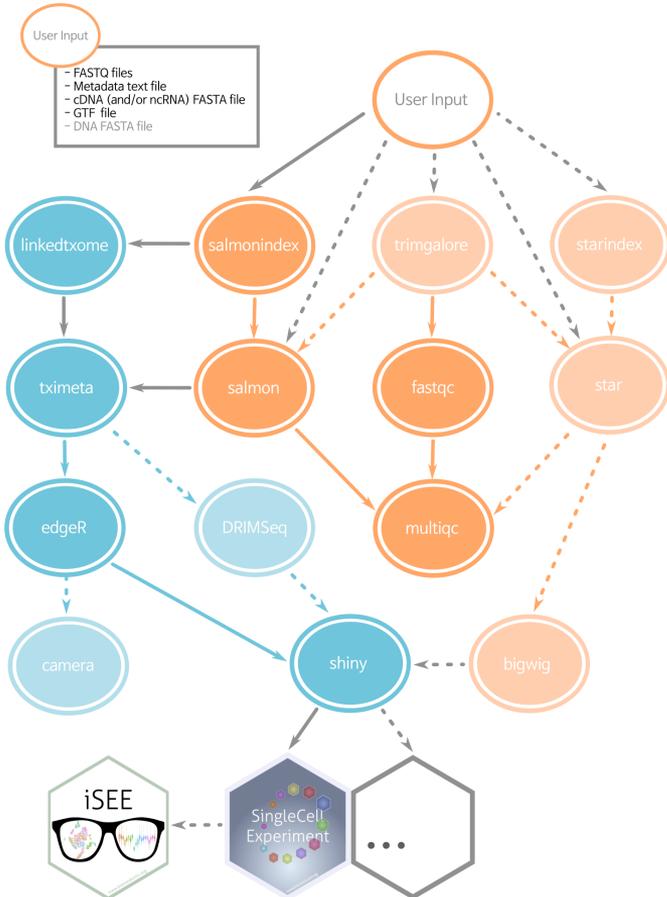
Autism spectrum disorder



Sibley et al. Nature Reviews 2016

Downregulation of **microexons** (≤ 27 nts).

ARMOR (Automated Reproducible MOdular RNA-seq)

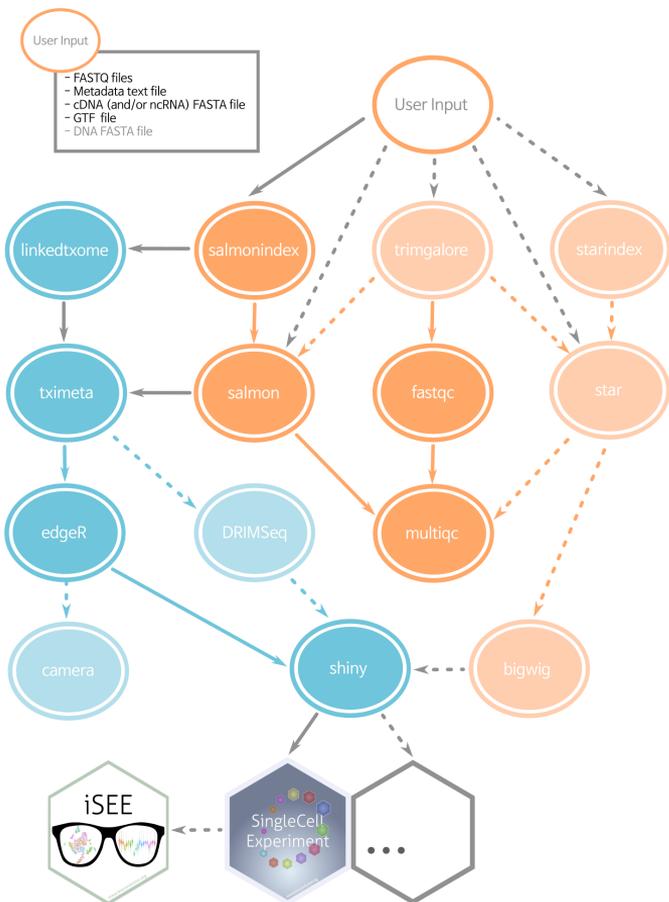


- RNA-seq analysis pipeline
- snakemake workflow



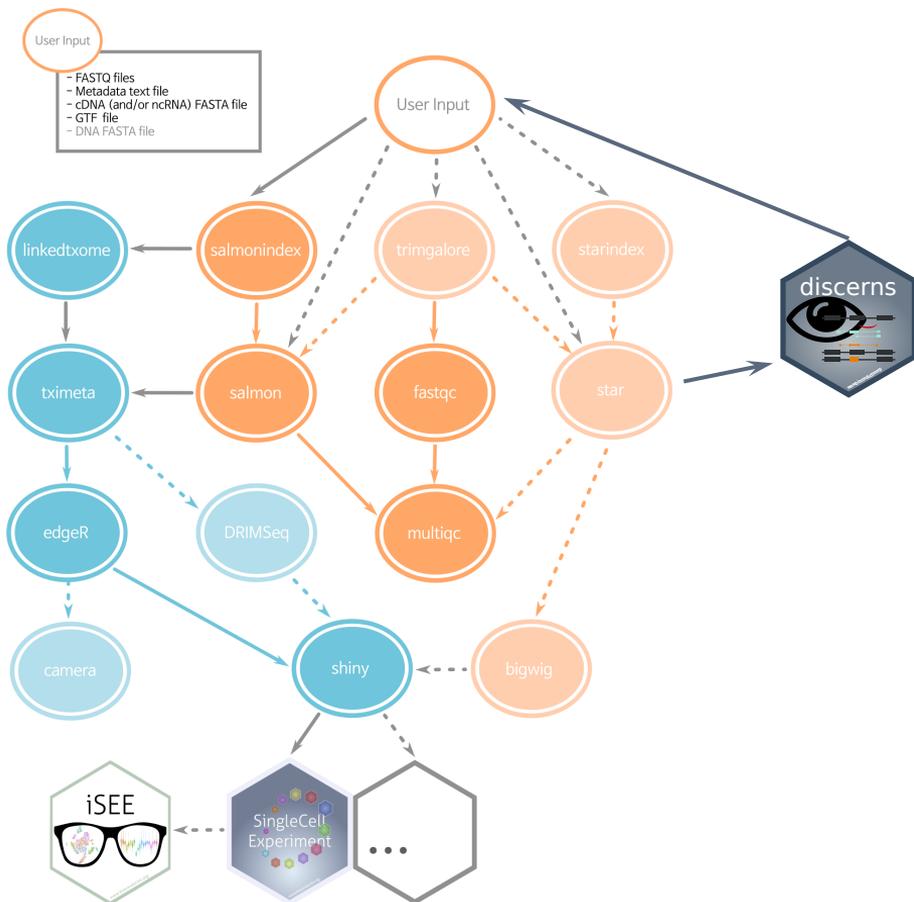
<https://github.com/csoneson/ARMOR>

Where does **discerns** fit into ARMOR?



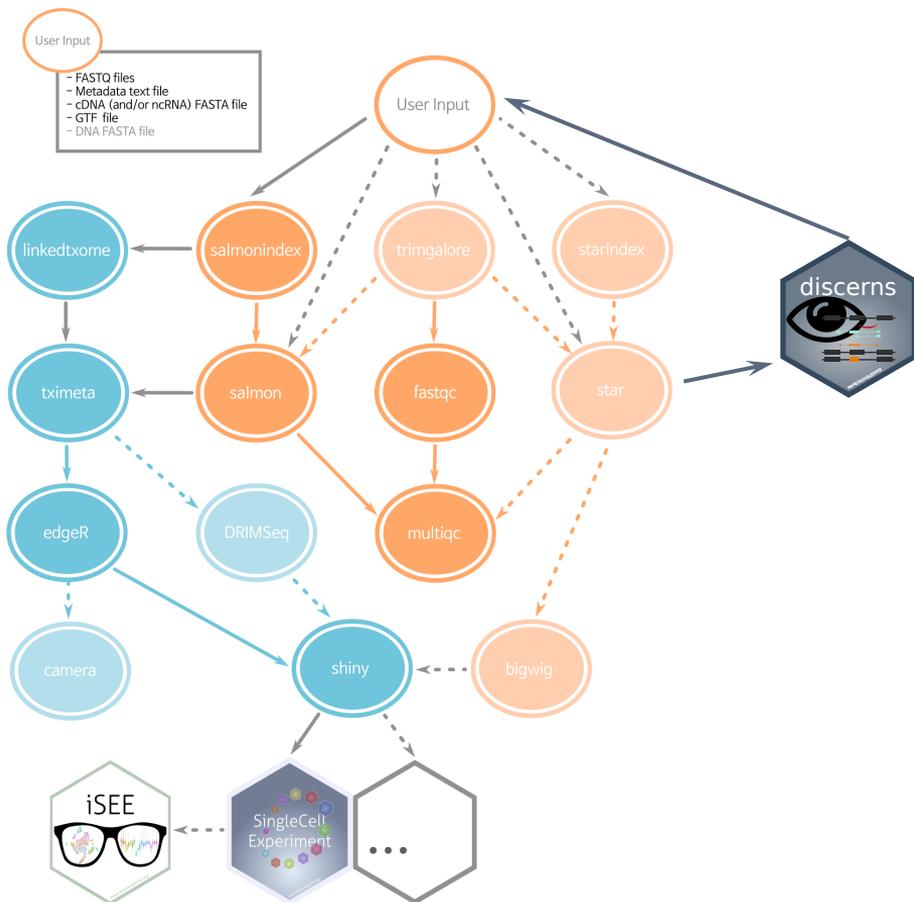
① **align** reads with STAR (Dobin et al. 2013)

Where does **discerns** fit into ARMOR?



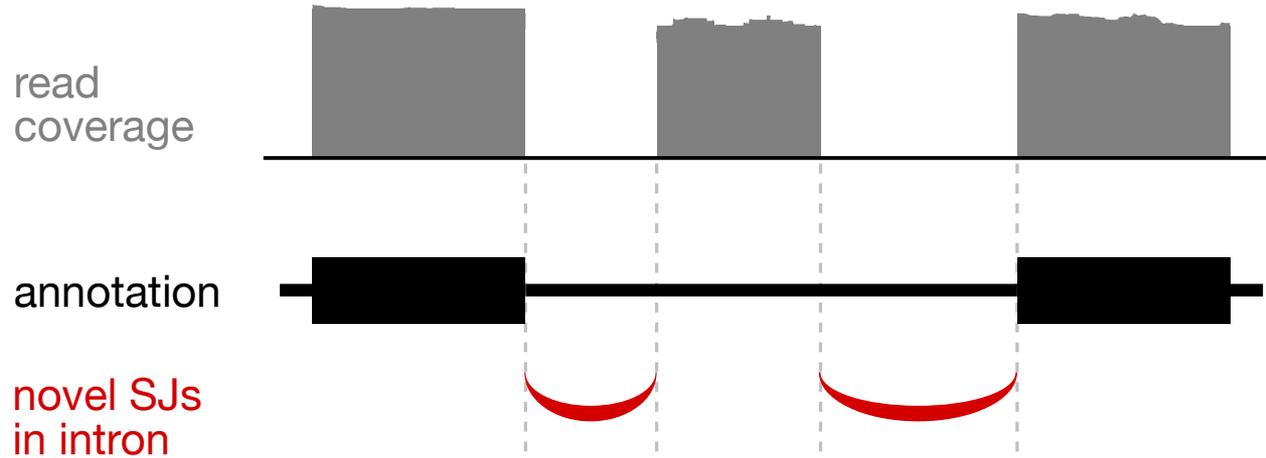
- ① **align** reads with STAR (Dobin et al. 2013)
- ② **predict** novel exons genome-wide (**discerns**)
- ③ **extend** annotation with novel exons (**discerns**)

Where does **discerns** fit into ARMOR?

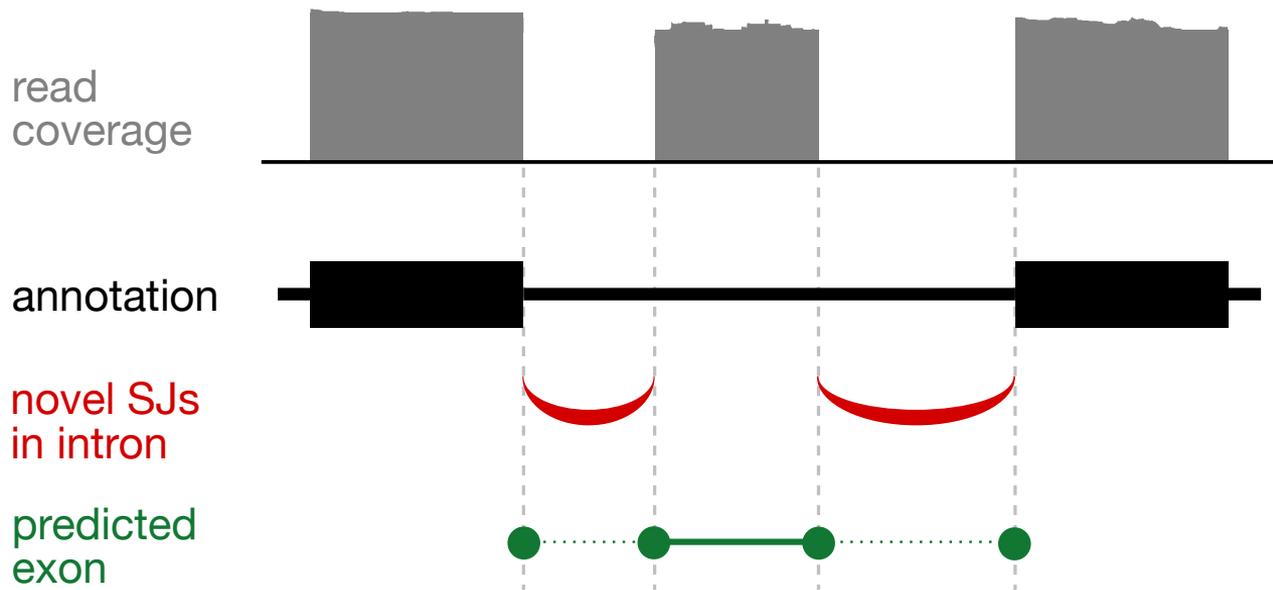


- ① **align** reads with STAR (Dobin et al. 2013)
- ② **predict** novel exons genome-wide (**discerns**)
- ③ **extend** annotation with novel exons (**discerns**)
- ④ **compare** predictions of samples with different treatment, genotype,...

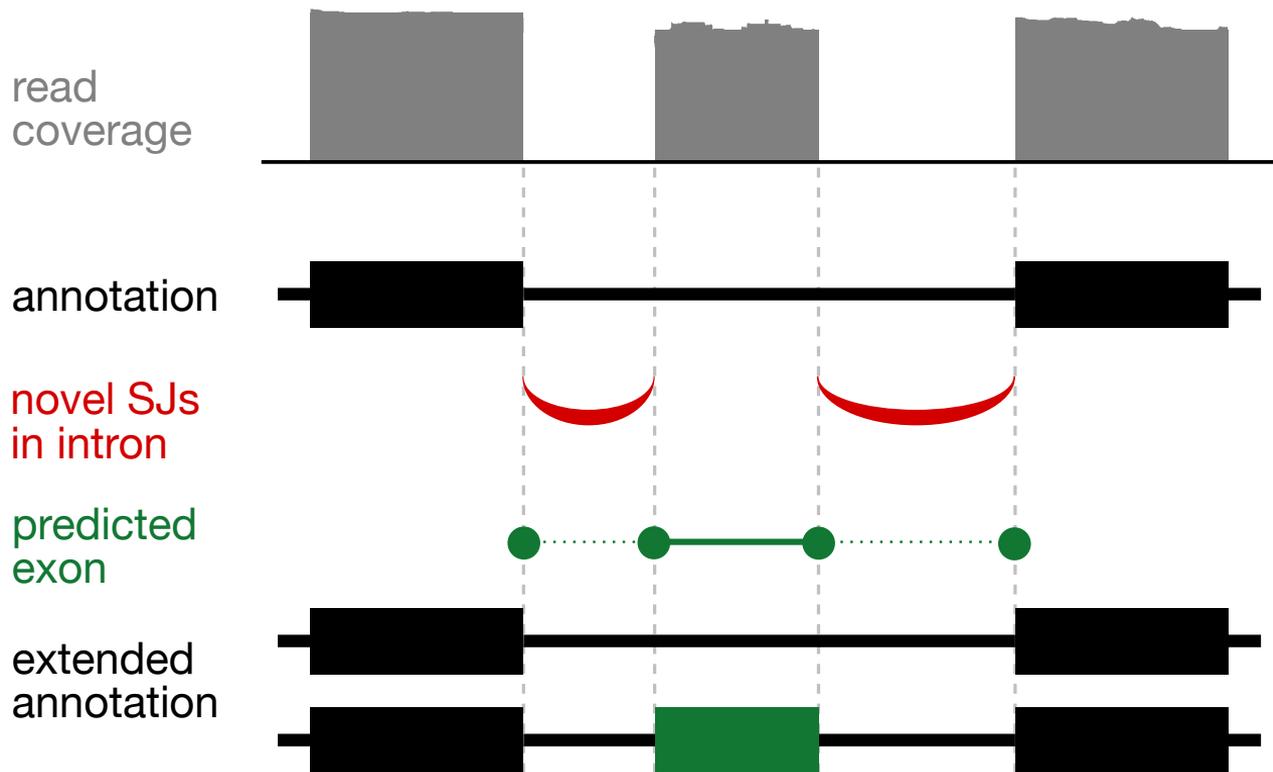
discerns exon prediction: 2 novel splice junctions



discerns exon prediction: 2 novel splice junctions



discerns exon prediction: 2 novel splice junctions



discerns exon prediction: 1 novel splice junction

internal exons

read coverage



annotation

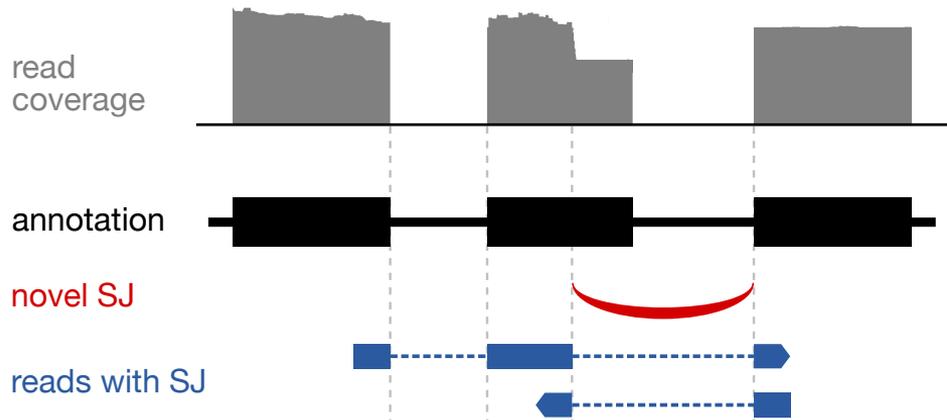


novel SJ



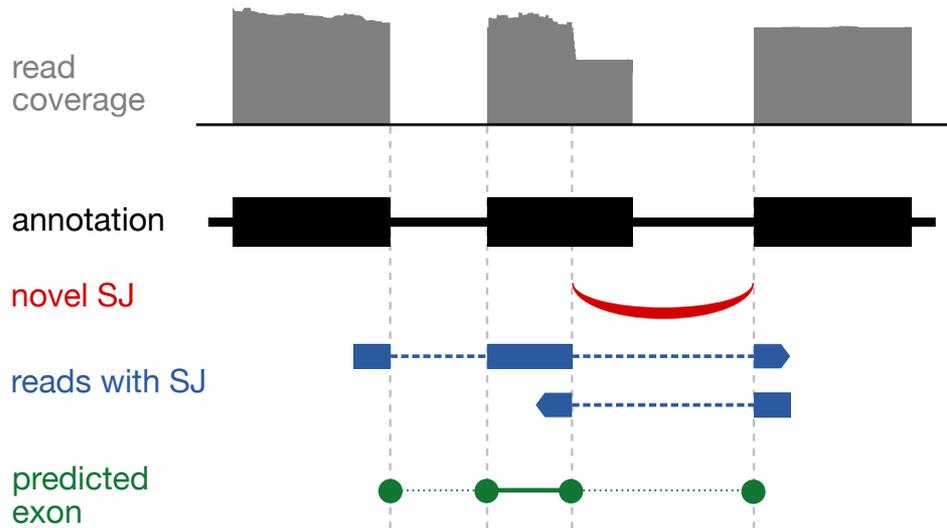
discerns exon prediction: 1 novel splice junction

internal exons



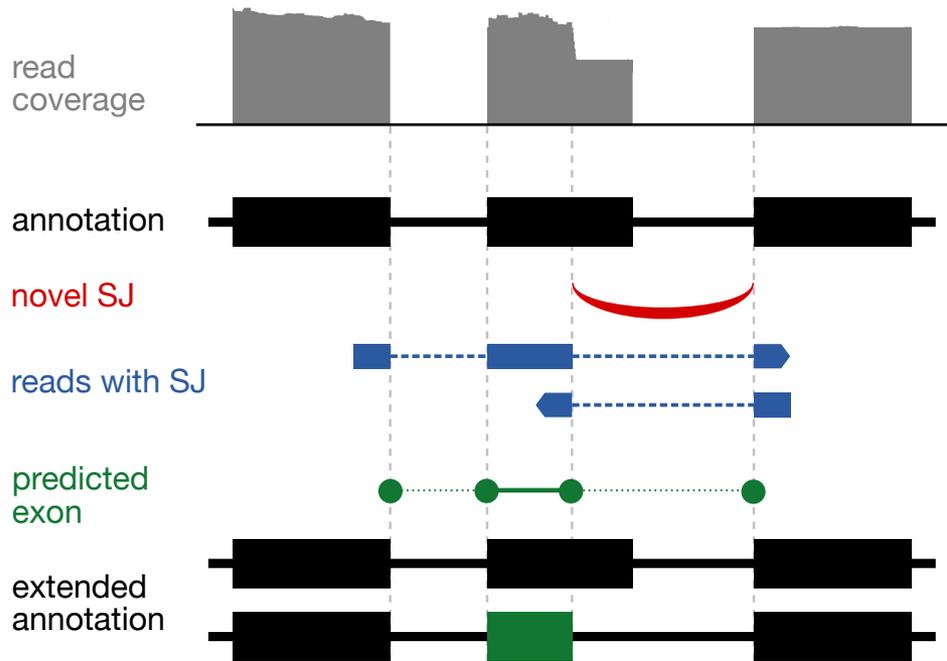
discerns exon prediction: 1 novel splice junction

internal exons



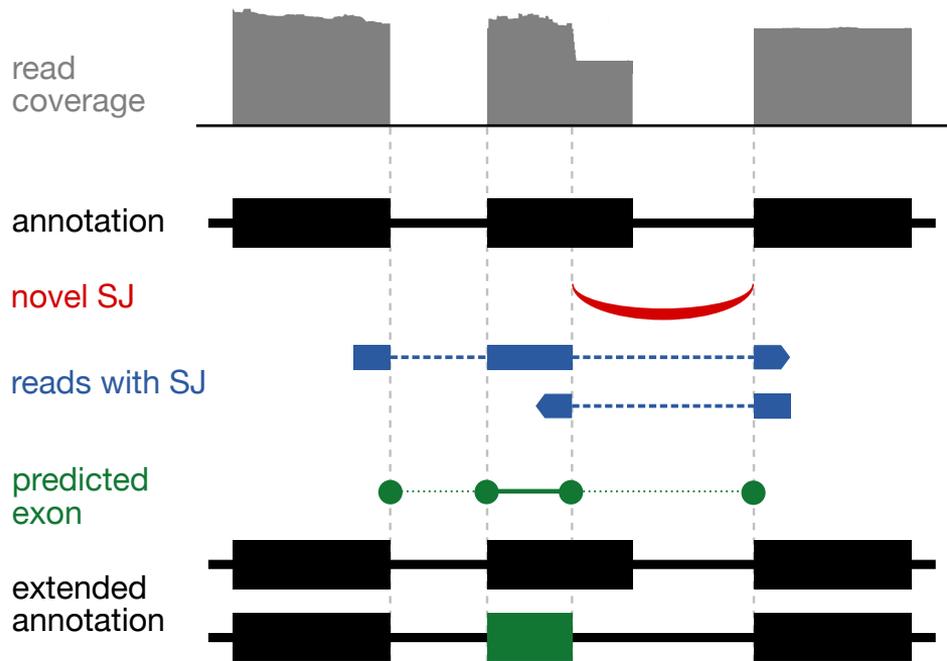
discerns exon prediction: 1 novel splice junction

internal exons

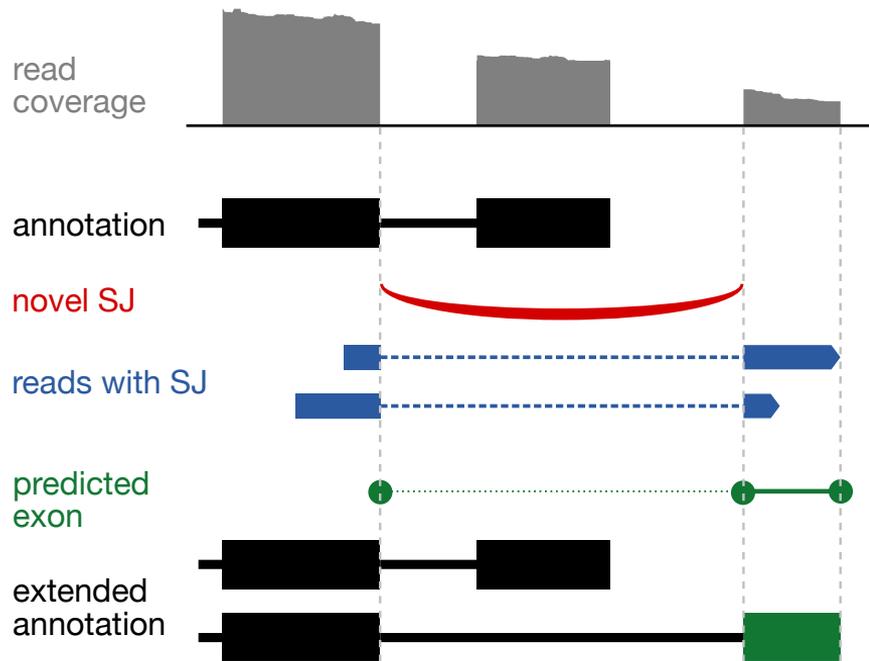


discerns exon prediction: 1 novel splice junction

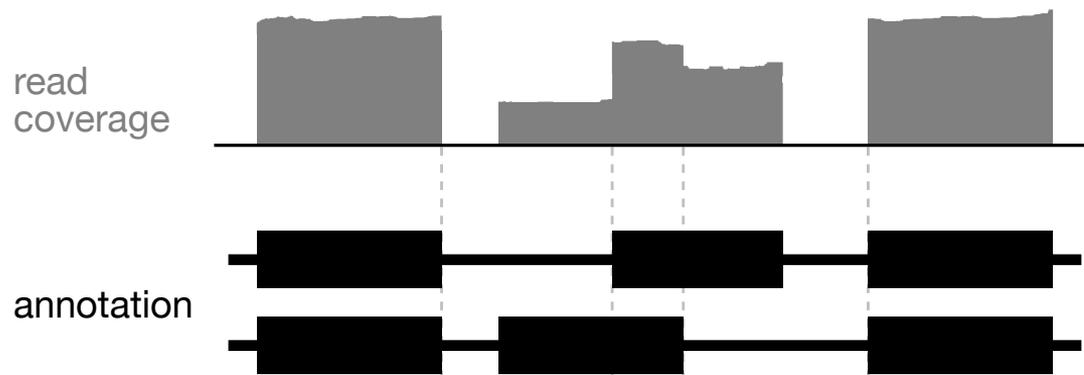
internal exons



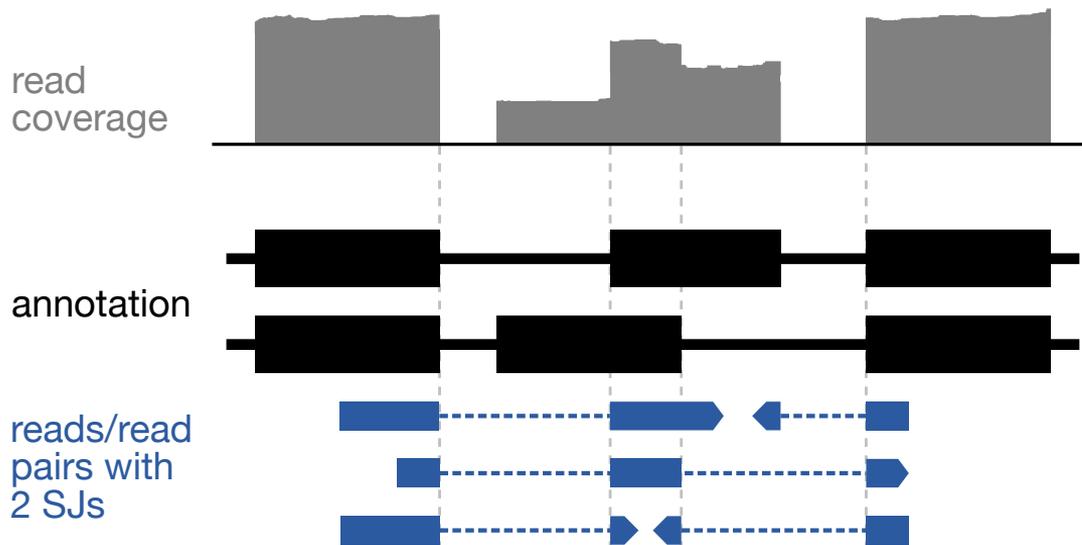
terminal exons



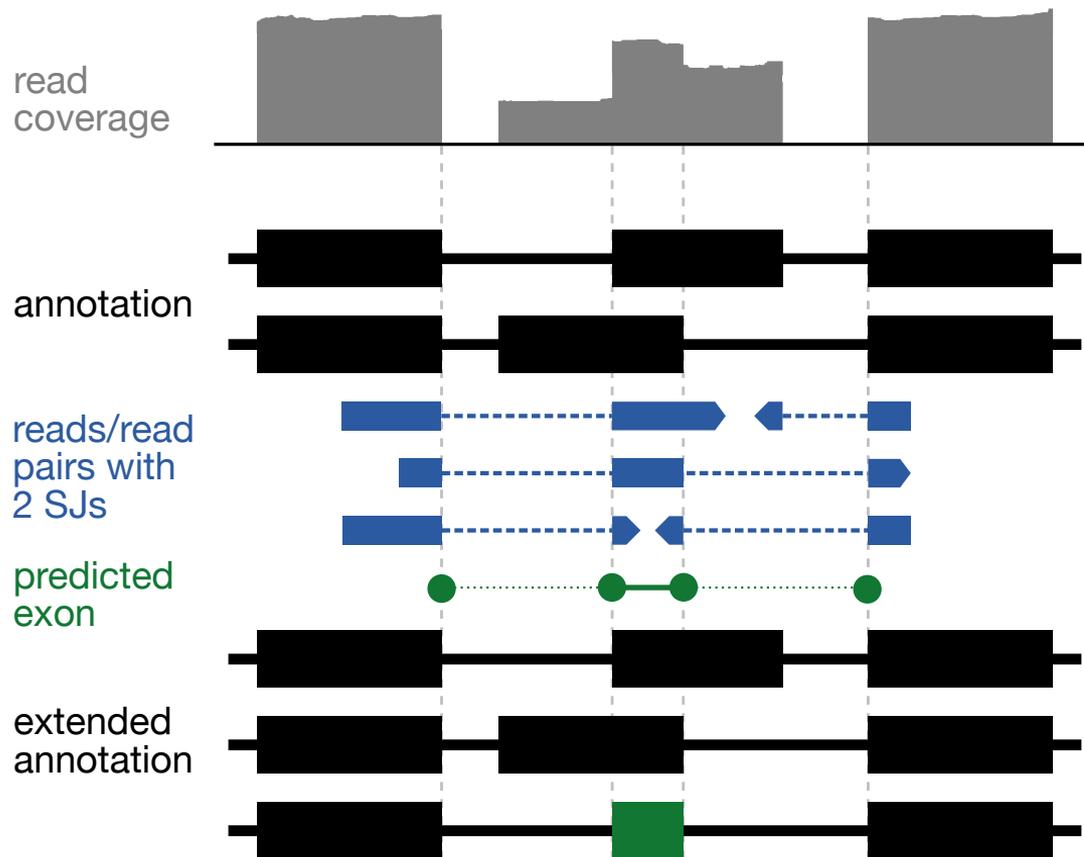
discerns exon prediction: 0 novel splice junction



discerns exon prediction: 0 novel splice junction



discerns exon prediction: 0 novel splice junction



discerns main functions

```
find_novel_exons(  
sj_filename,  
annotation, bam,  
min_unique = 1,  
cores = 1, ...)
```

- ▣ **Input:** SJ.out.tab from STAR, annotation object, BAM file
- ▣ **Output:** data.frame with coordinates of novel exons and # supporting reads

discerns main functions

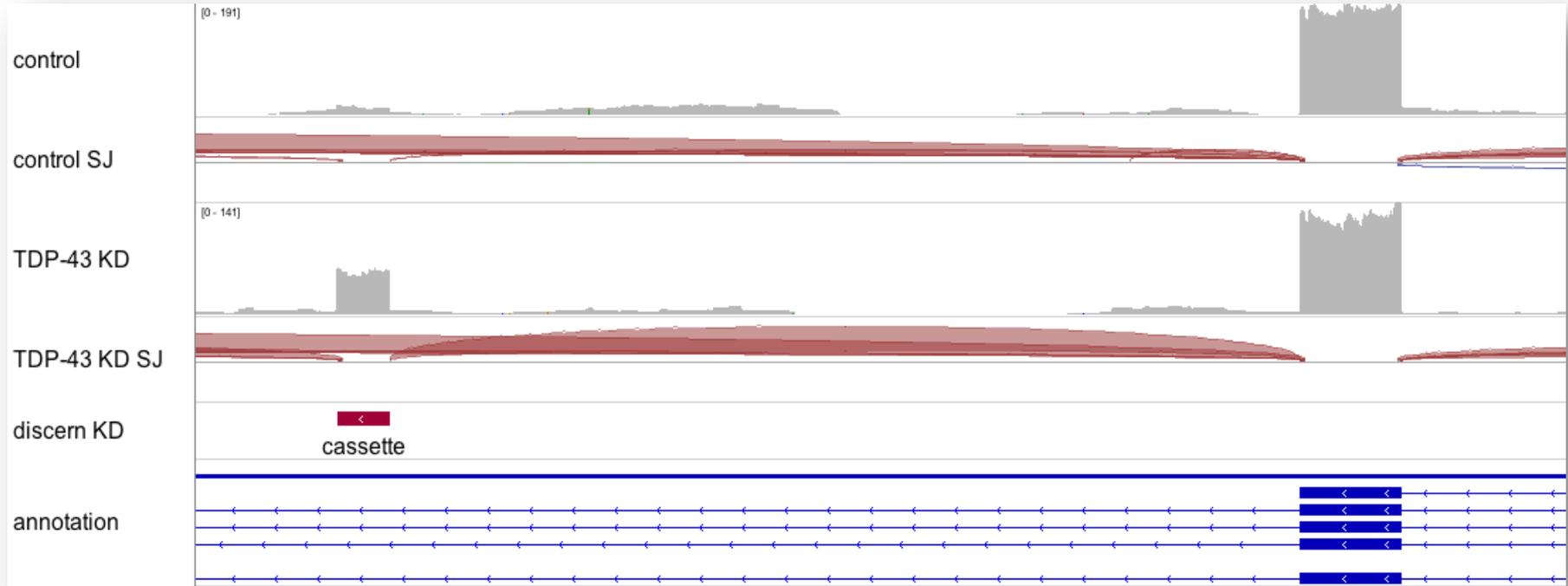
```
pred <-  
find_novel_exons(  
  sj_filename,  
  annotation, bam,  
  min_unique = 1,  
  cores = 1, ...)
```

- ▣ **Input:** SJ.out.tab from STAR, annotation object, BAM file
- ▣ **Output:** data.frame with coordinates of novel exons and # supporting reads

```
extend_gtf(  
  gtf,  
  pred,  
  cores = 1)
```

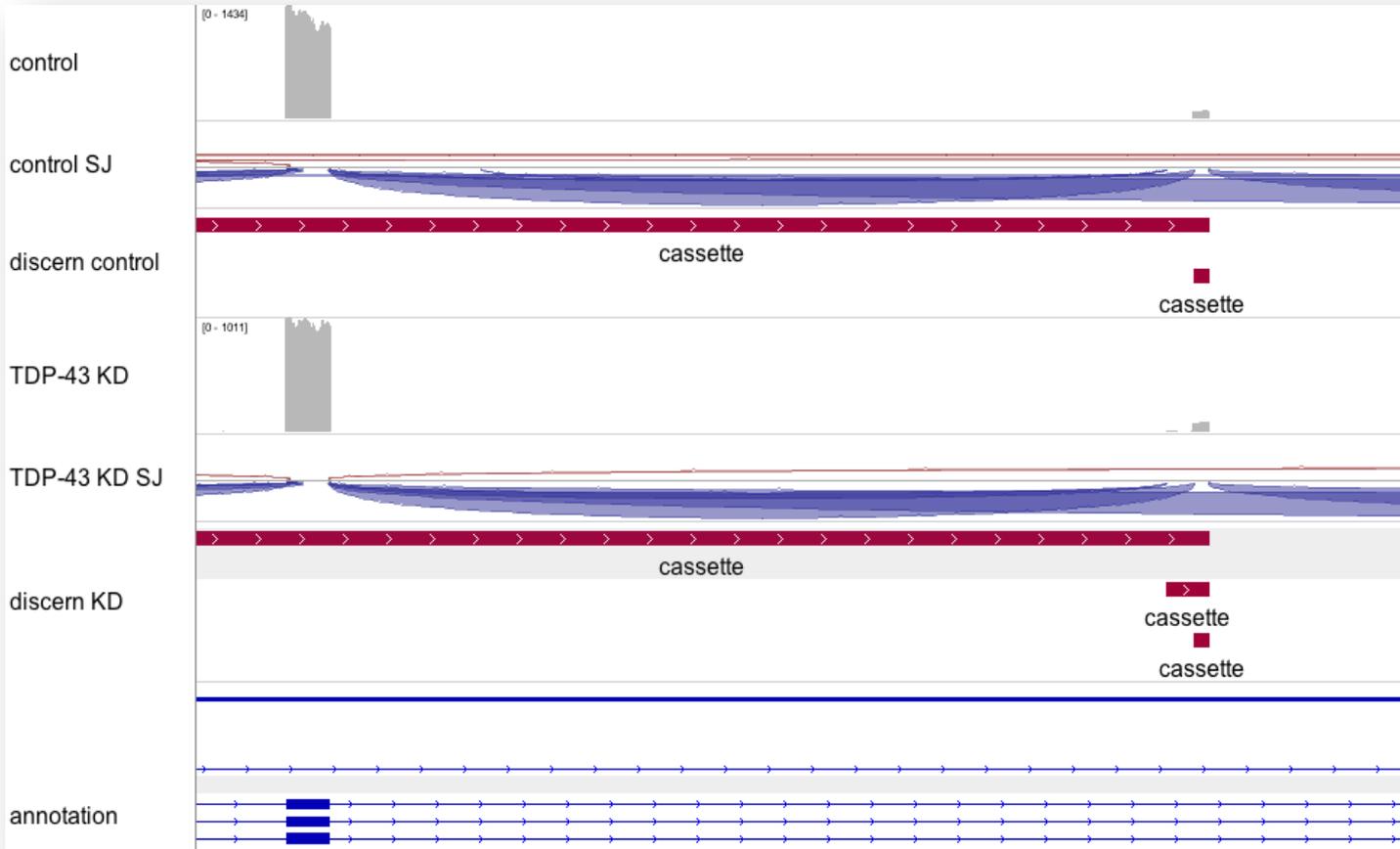
- ▣ **Input:** GTF file, predicted exons
- ▣ **Output:** GTF annotation with new transcript per novel exon

discerns identifies novel exons in real RNA-seq data

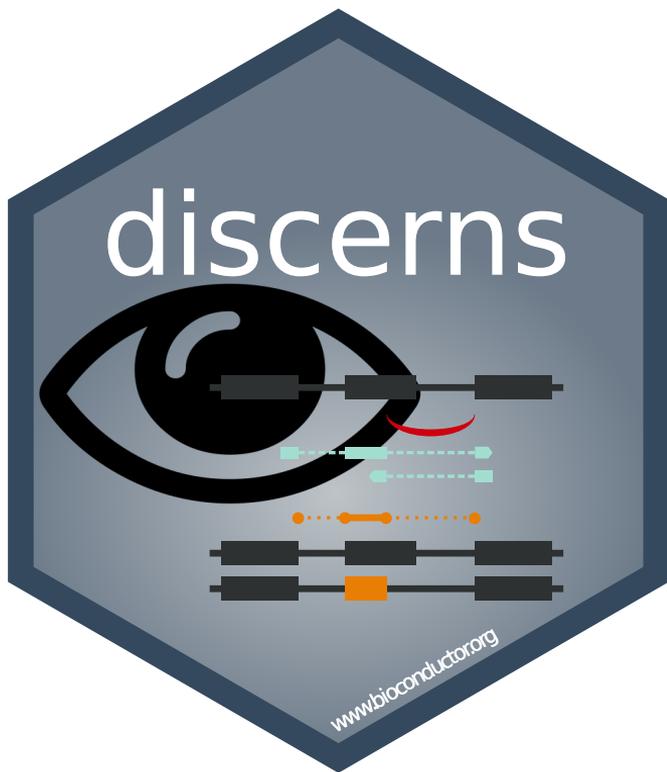


cryptic exon

discerns identifies novel exons in real data



microexon



will soon be submitted to
Bioconductor



<https://github.com/csoneson/ARMOR>