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NBIS NATIONAL BIOINFORMATICS INFRASTRUCTURE SWEDEN

Jacques Dainat PhD

The genome annotation pipeline

MAKER

Annotate this!

elixir

excelerate

Uppsala – May 2019

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Generic Model Organism Database project

GMOD in the Cloud toolset

GBrowse: Genome annotation viewer

GMOD WORLD

Galaxy: Data analysis & integration

Chado: Biological database schema

JBrowse: Super-fast genome annotation viewer

BioMart: Data mining system

WebApollo: browser-based annotation editor

MAKER: Genome annotation pipeline

GBrowse_syn: Synteny viewer

Tripal: Chado web interface

InterMine: Data warehousing

CMap: Comparative map viewer

Pathway Tools: Metabolic, regulatory pathways

Canto: literature annotation tool

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Eukaryotic genome annotation

MAKER: An easy-to-use annotation pipeline designed for ... - NCBI - NIH
https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2134774/ ▾ Översätt den här sidan
av BL Cantarel - 2008 - Citerat av 612 - Relaterade artiklar
We have developed a portable and easily configurable genome **annotation pipeline** called **MAKER**. Its purpose is to allow investigators to independently ...
Abstract · Results · Discussion · Methods

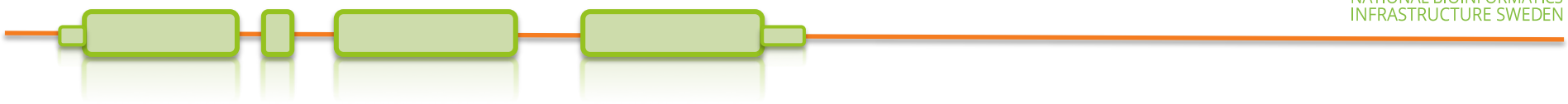
09/2018

MAKER2: an annotation pipeline and genome ... - BMC Bioinformatics
https://bmcbioinformatics.biomedcentral.com/.../1471-2105-12-... ▾ Översätt den här sidan
av C Holt - 2011 - Citerat av 514 - Relaterade artiklar
22 dec. 2011 - We present **MAKER2**, a genome **annotation** and data management tool designed for second-generation genome projects. **MAKER2** is a ...

MAKER – developed as an easy-to-use alternative to other pipelines

Why choose MAKER ?

- Easy to use and to configure
- Scale to datasets of any size
- Multi-threaded and parallelized
- Everything is run through one command, no manual combining of data/outputs
- Metric for quality control (**A**nnotation **E**dit **D**istance)
- Distributed with accessory scripts (>30)
- All its capabilities…



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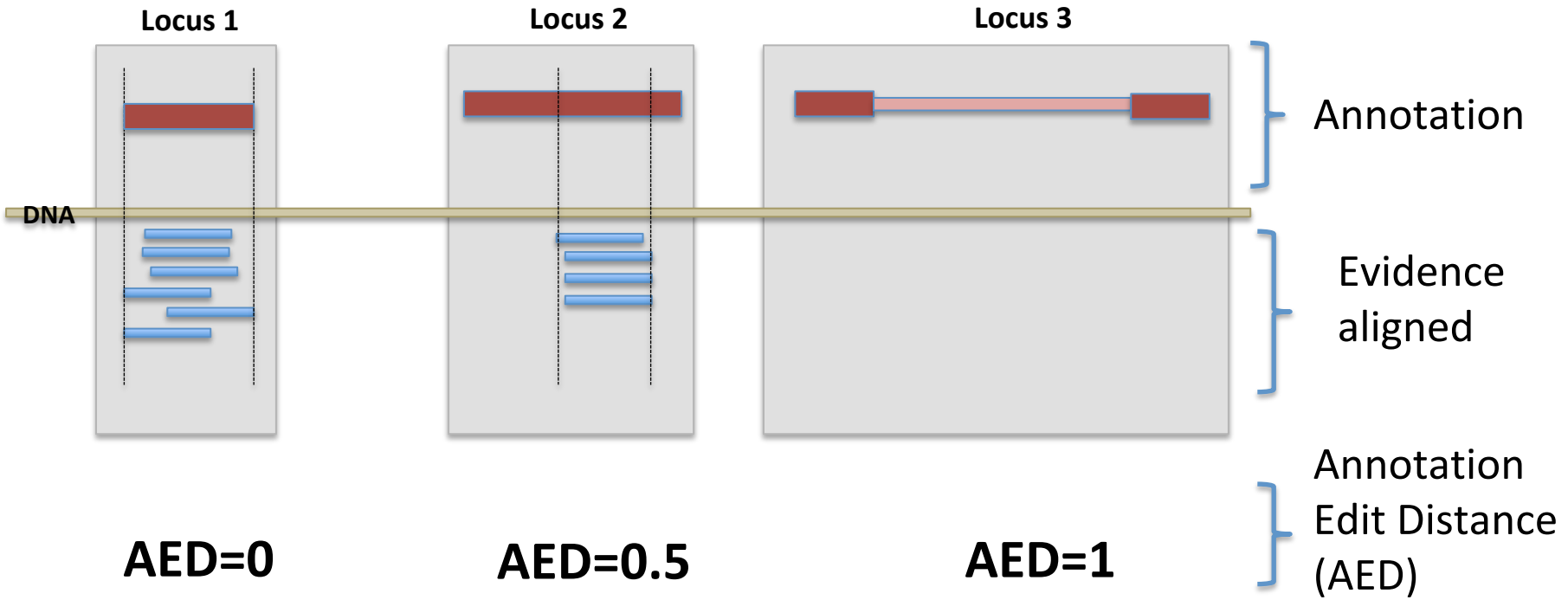
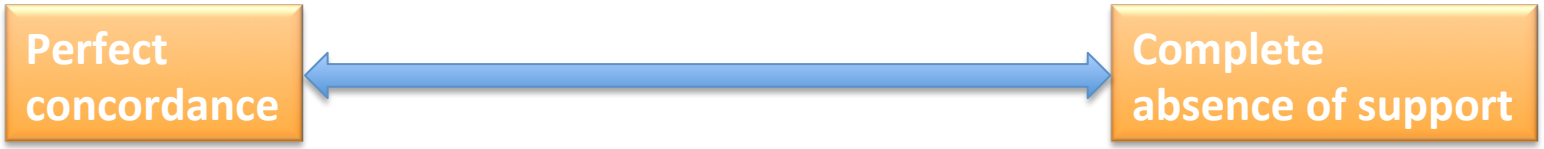
Capabilities:

- ❑ Add quality metrics to an annotation (AED)





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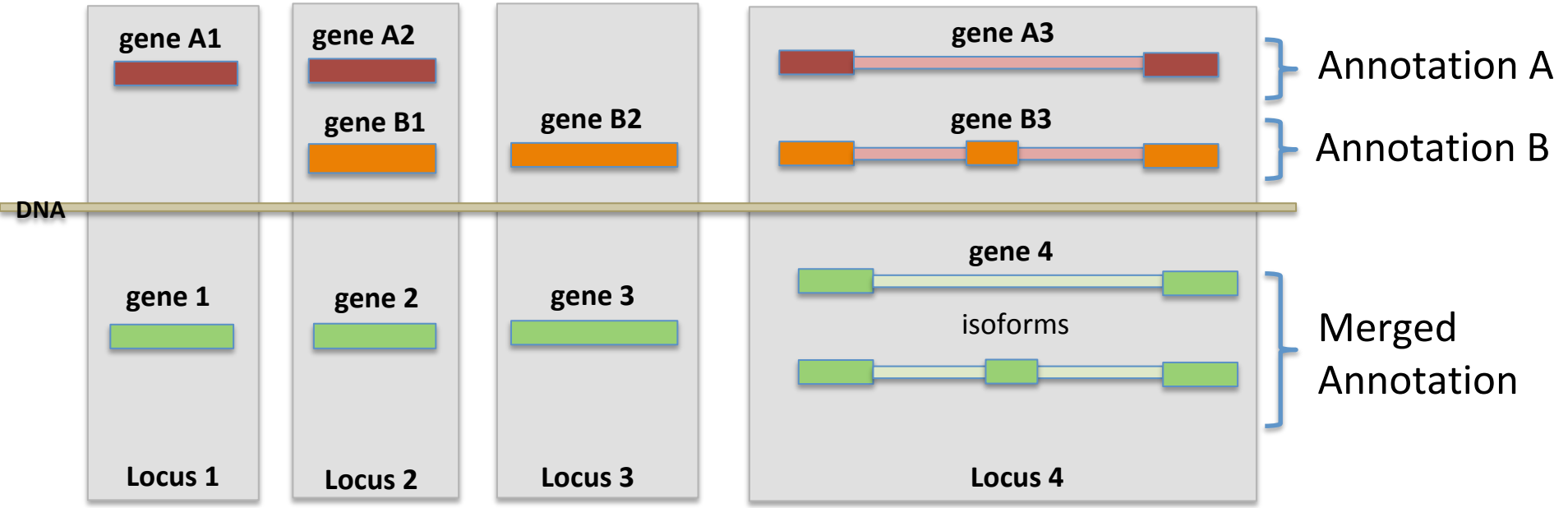
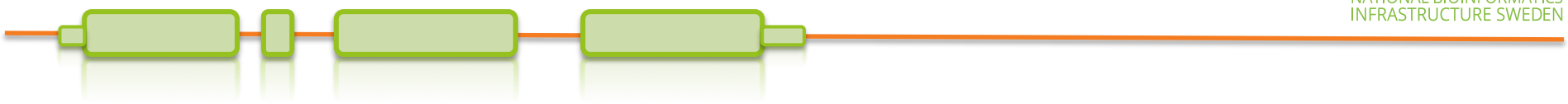
Capabilities:

- ❑ Merge multiple annotation sets





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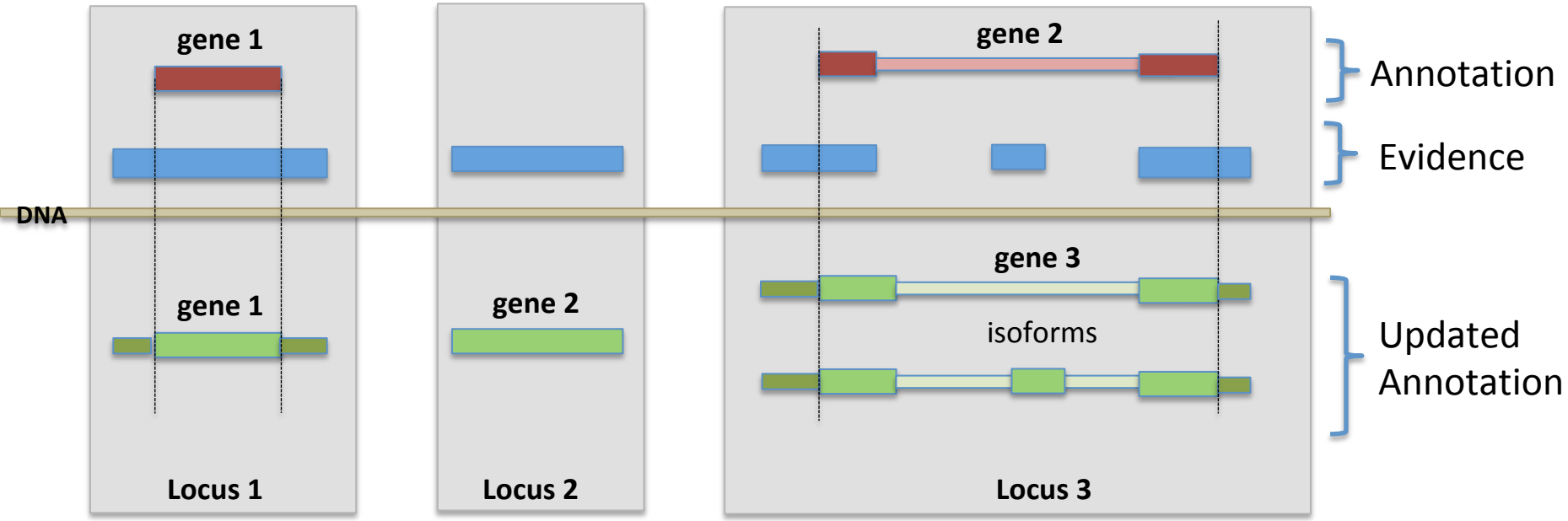
Capabilities:

- ❑ Update an annotation in light of new evidence



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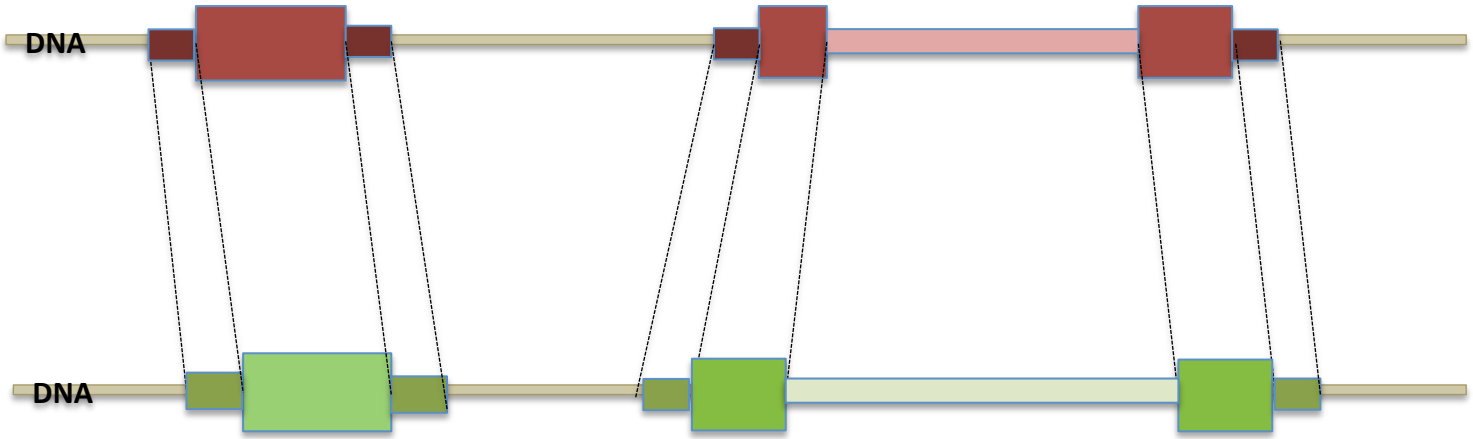
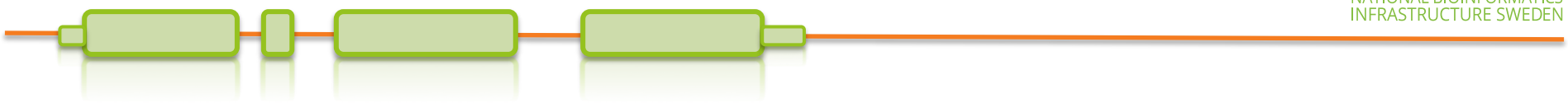
Capabilities:

- ❑ Map annotation forwards to a new assembly



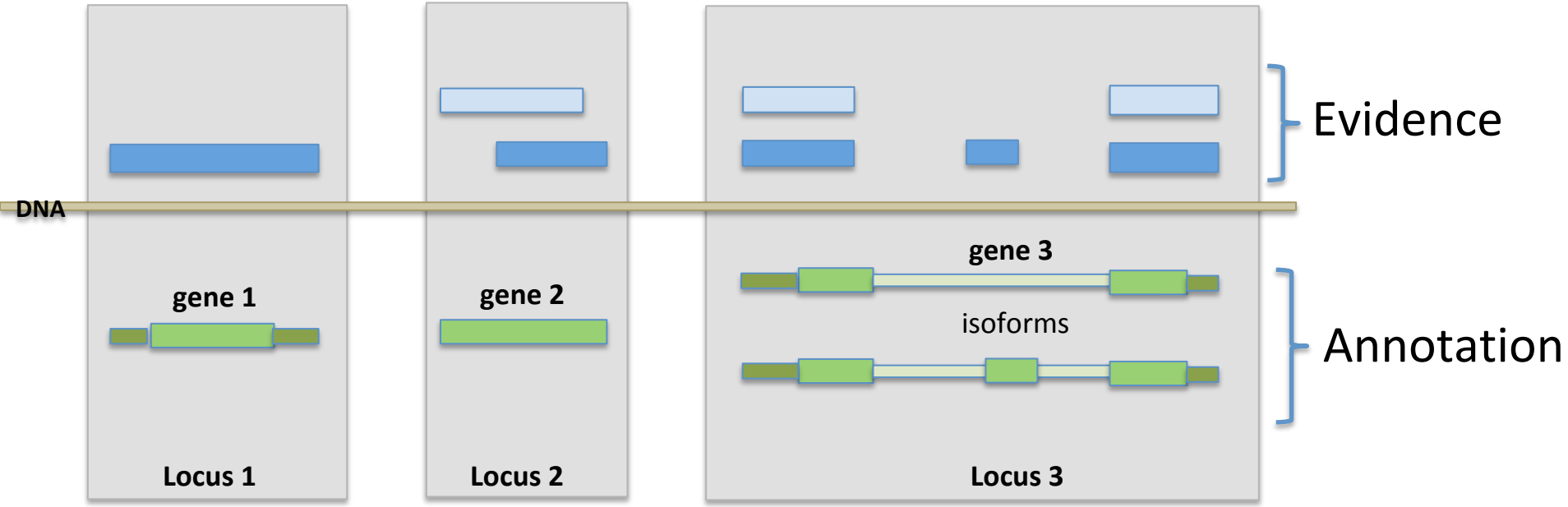
Annotation Assembly 1

Lift-over on Assembly 2



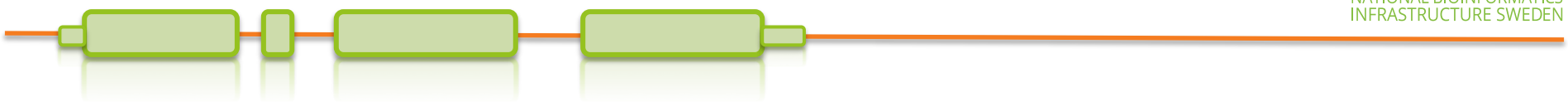
Capabilities:

- ❑ Annotation pure evidence-based



⇒ Suitable for ab-initio training purpose (filtering needed)

⇒ Prediction not always complete ! (always_complete option)



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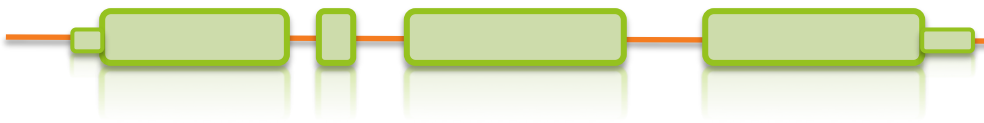
Capabilities:

- ❑ Annotation pure *ab-initio*

The same as standalone *ab-initio* ! So why MAKER ?

⇒ To take advantage of parallelization !

⇒ Can use several *ab-initio* tools (they can complement each other)



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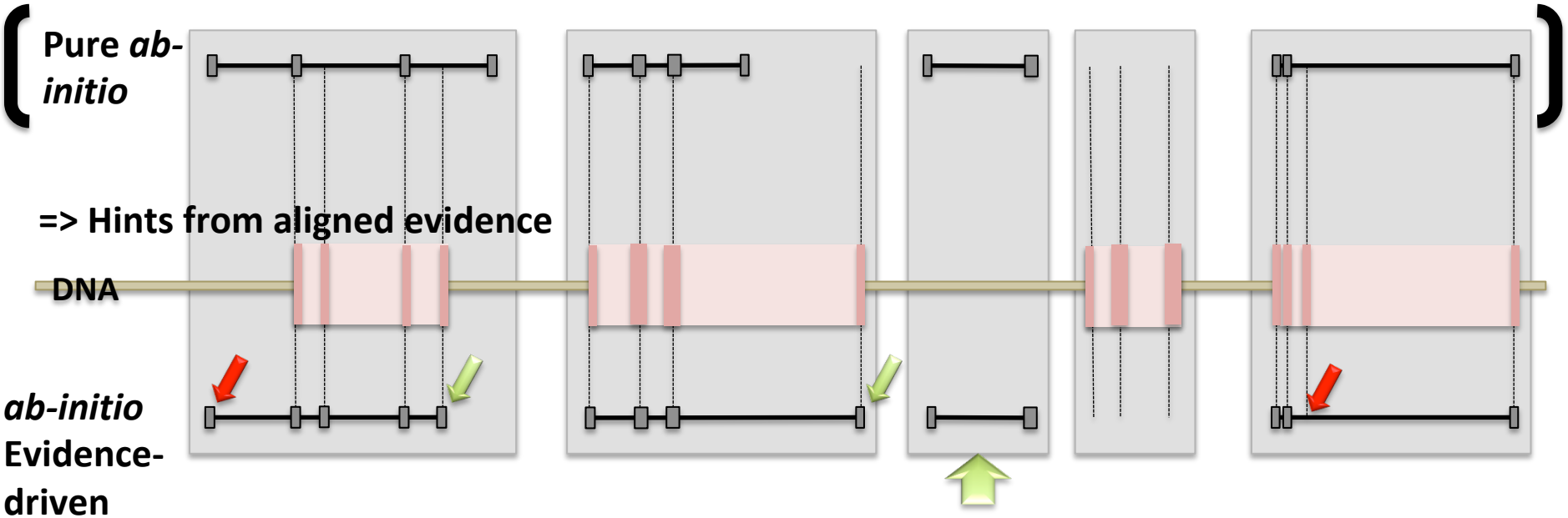
Capabilities:

- ❑ Annotation *ab initio* evidence-driven



Ab-initio tools are better when hints are provided

Ab-initio predictions can fill gaps with no evidence

⇒ But may still be incomplete / partially wrong

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But how does Maker work exactly?

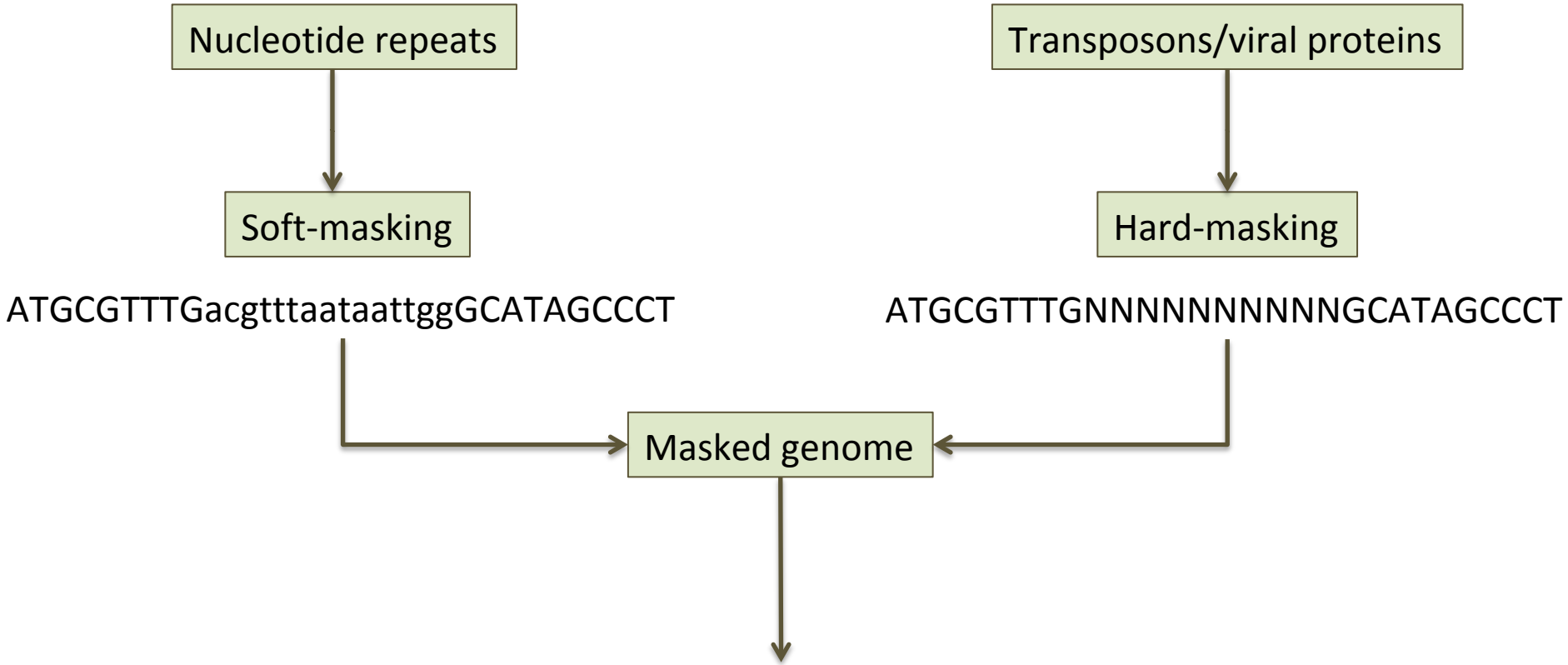
Use case

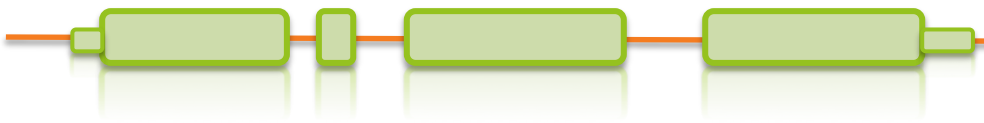
⇒*Ab-initio* evidence-driven

Prerequisite:

- ❑ Evidence (proteins and/or transcripts)
- ❑ Hmm profile for *ab-initio* tool(s)
  (Augustus comes with some pre-calculated profiles)

Step 1 - Repeat masking with repeatmasker





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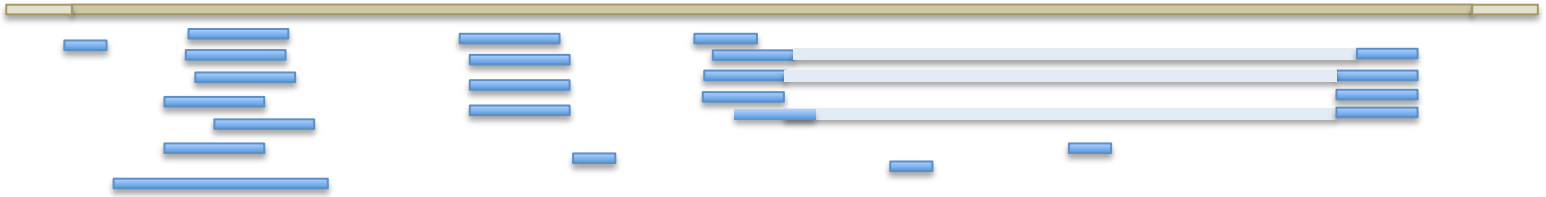
Step 2 - Transcript and protein evidence alignment

Step 3 - Filtering and clustering alignments



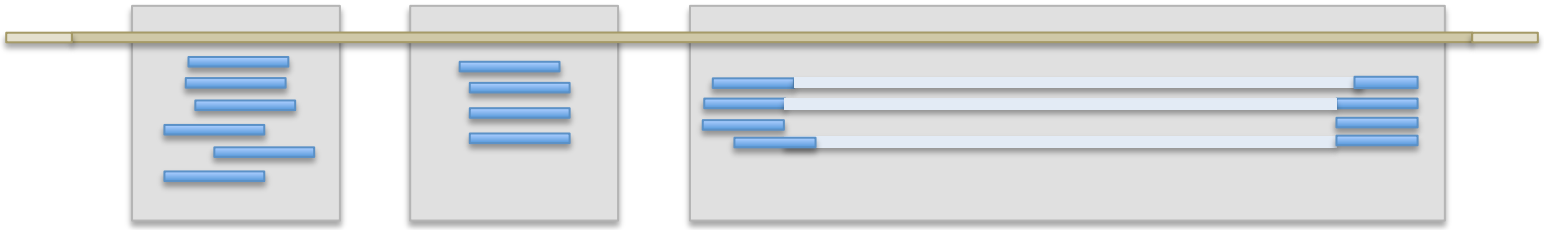
Filtering is based on rules defined in the Maker configuration for a given project

Example: EST alignment – 80% coverage and 85% identity

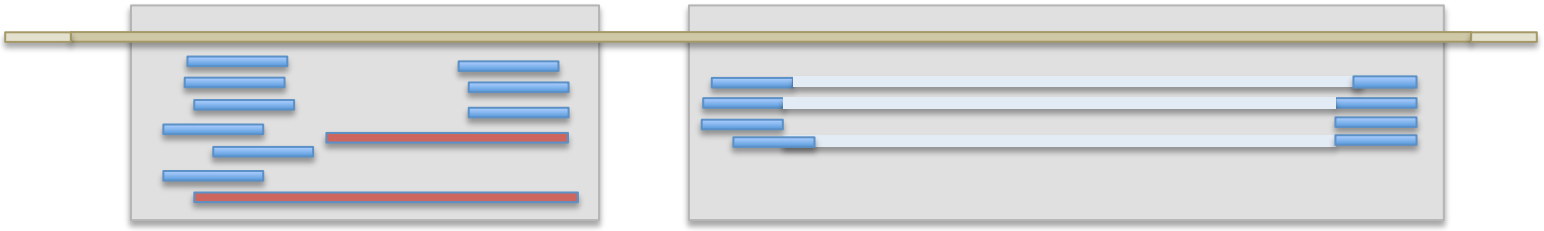
Default settings sensible for most projects, but can be changed!

Step 3 - Filtering and clustering alignments

Clustering into ’loci’

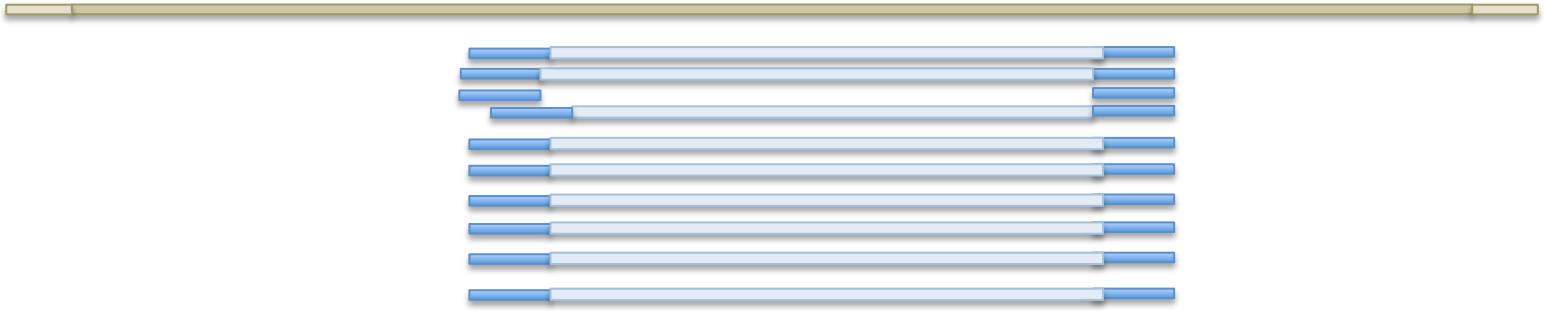


Importance of the quality of the data used:



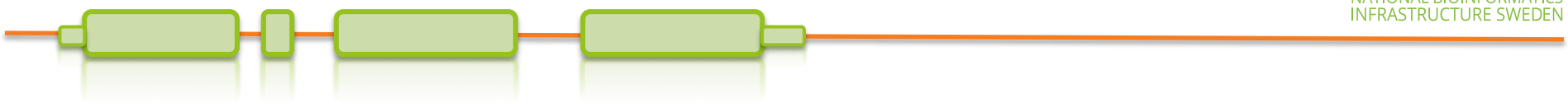
=> Bad data can complicate clustering

Step 3 - Filtering and clustering alignments



Amount of data in any given cluster is then collapsed to remove redundancy

Threshold for the collapsing is also user-definable



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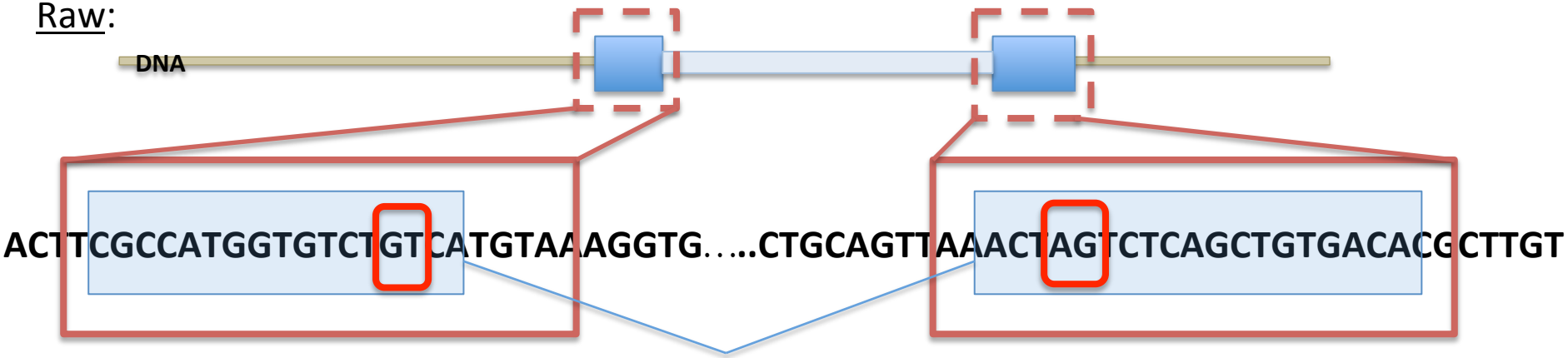
Step 4 - Polishing evidence alignments

Raw:



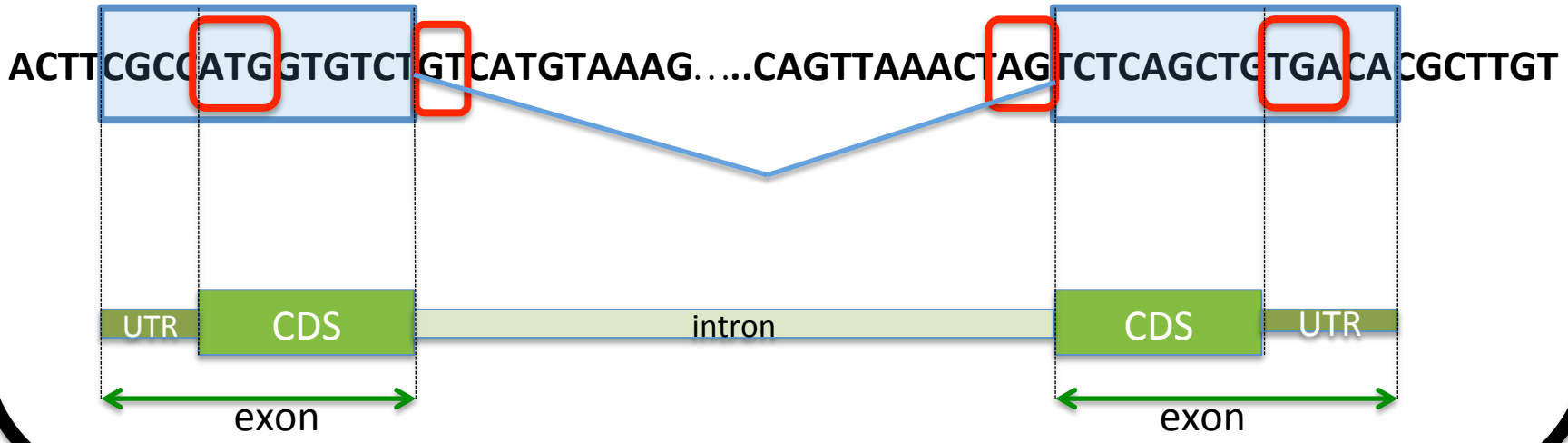
Blast-based alignments are only approximations, need to be refined

Exonerate is used to create splice-aware alignments

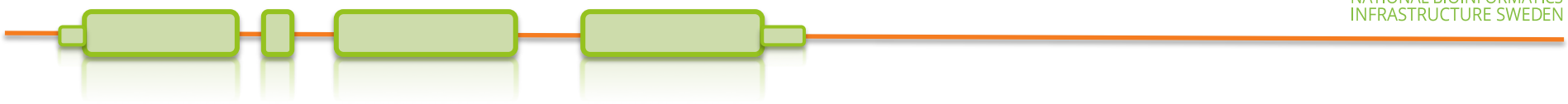
Polished:

Parenthesis !

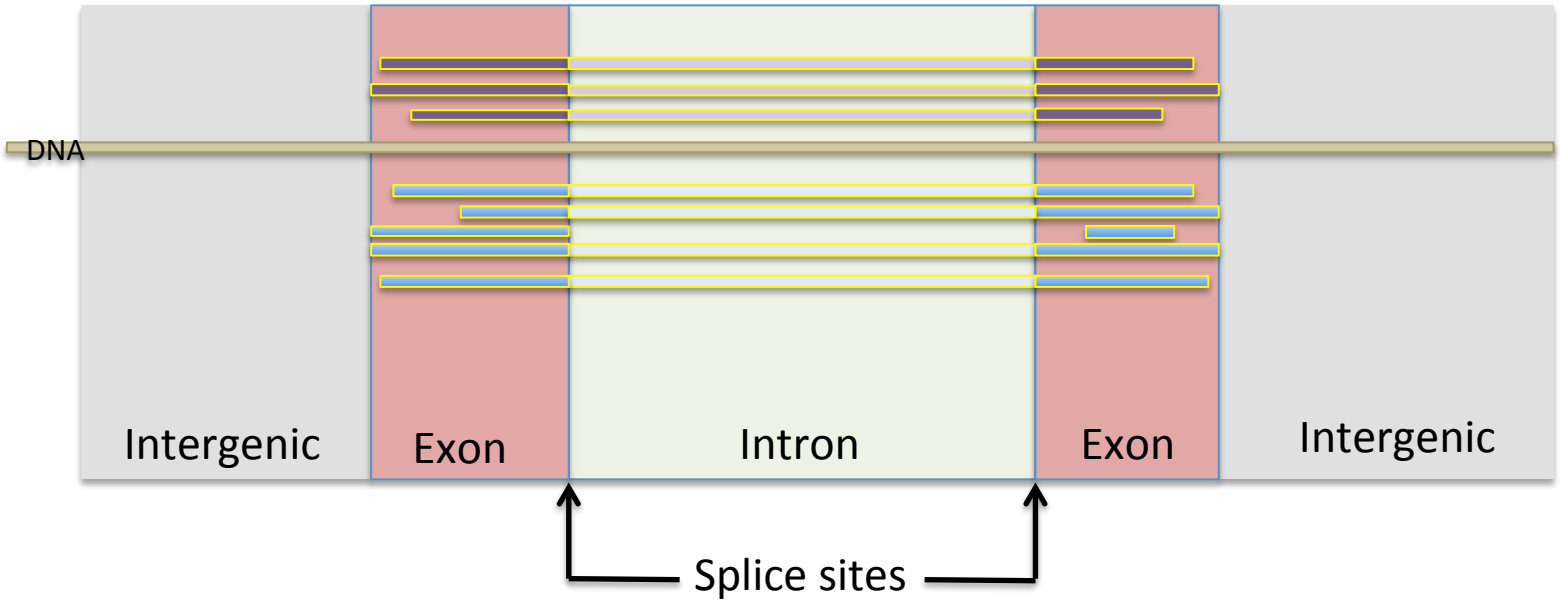
⇒ In a pure evidence based case, the last step will be the creation of gene model from polished alignments.



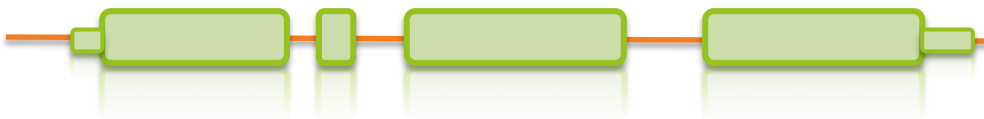
Let's get back on track !



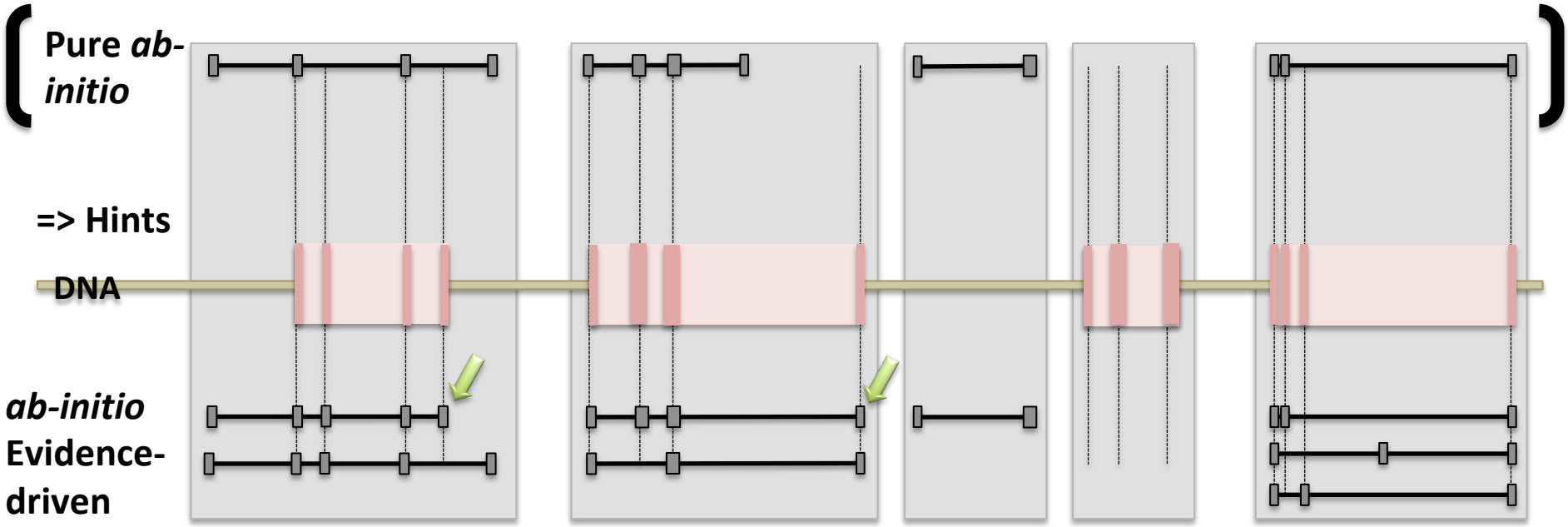
Step 5 – Generating hints



Hints are passed to *ab-initio* tools that accept them

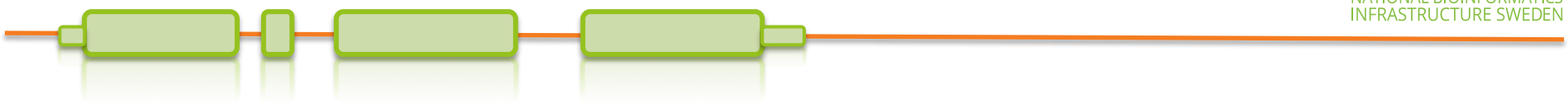


Step 6 - *Ab-initio* gene prediction (evidence-driven)

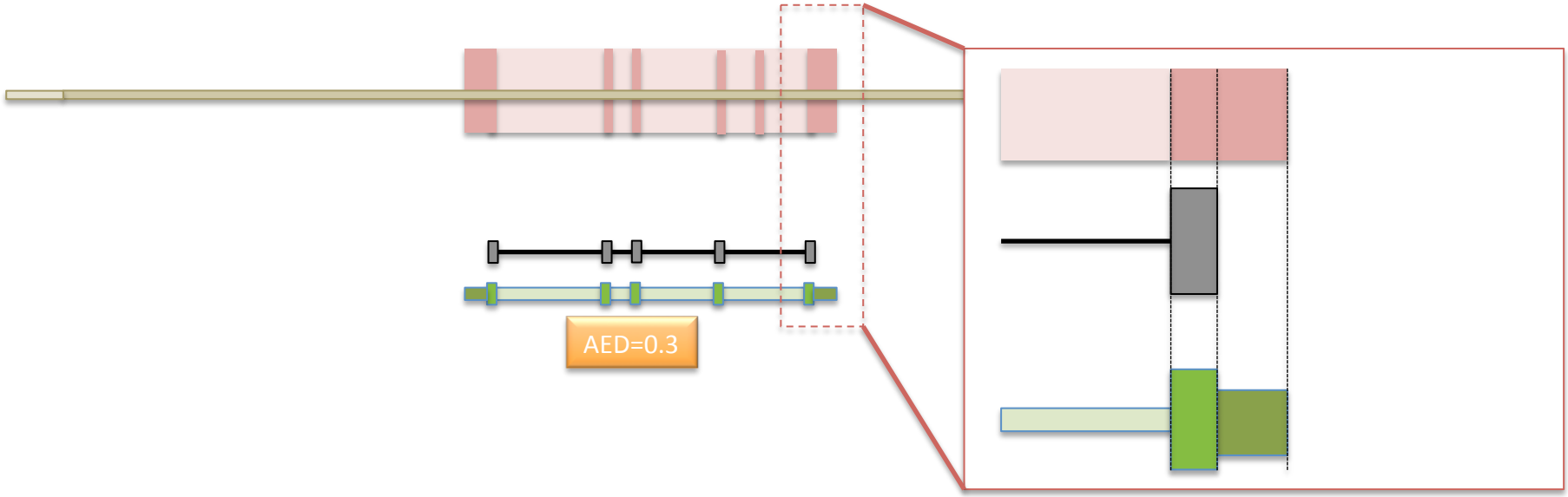


Ab-initio predictions are refined when hints are provided

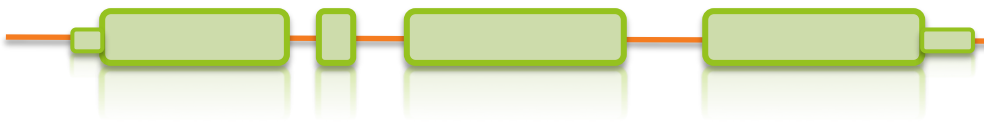
Isoforms accepted if parameter activated



Step 7 - Annotation

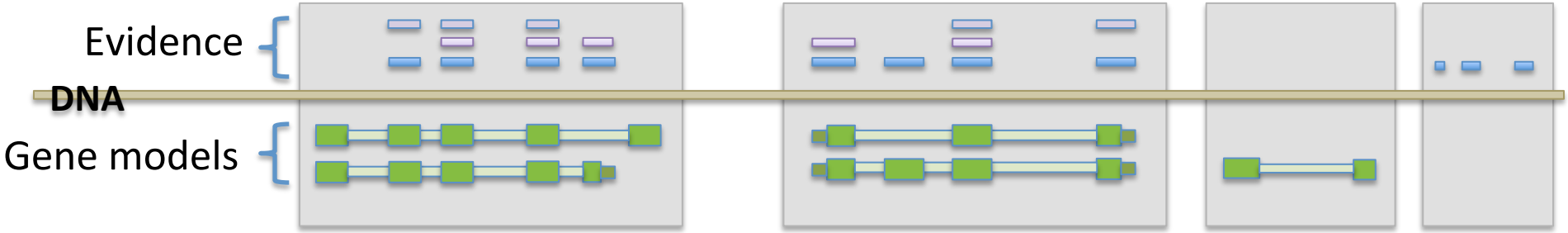


- Add UTRs
- Add quality control metrics



Step 8 – Selecting gene models

⇒ selected in agreement with the available evidence
⇒ The minimum agreement threshold can be chosen

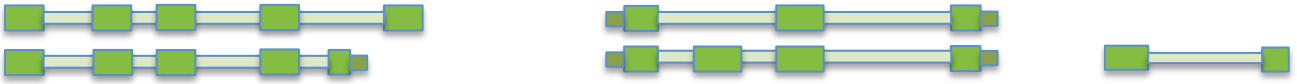


Final MAKER Annotation:

keep_preds=0
Maximum sensitivity

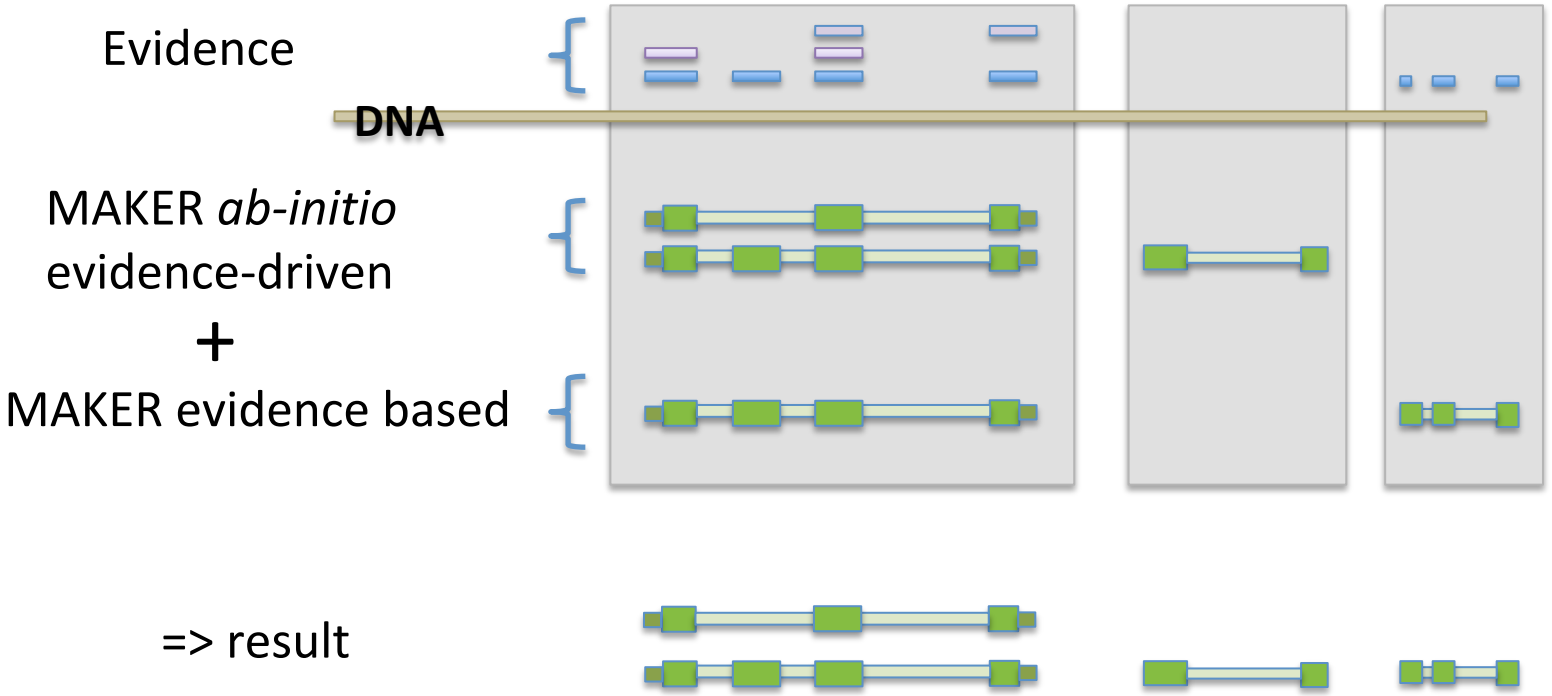
alt_splice=1

keep_preds=1



specificity ↑ sensitivity ↓

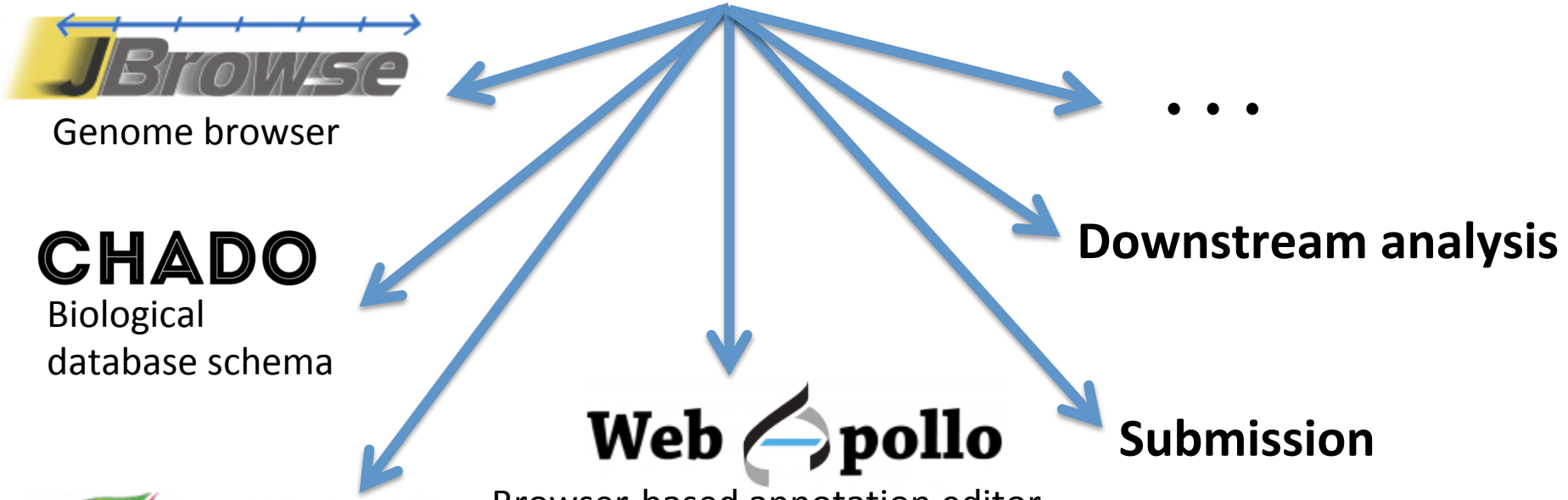
Tip - complete the annotation



What next ?

MAKER
Annotate this!

Output = Annotation in **gff3** format



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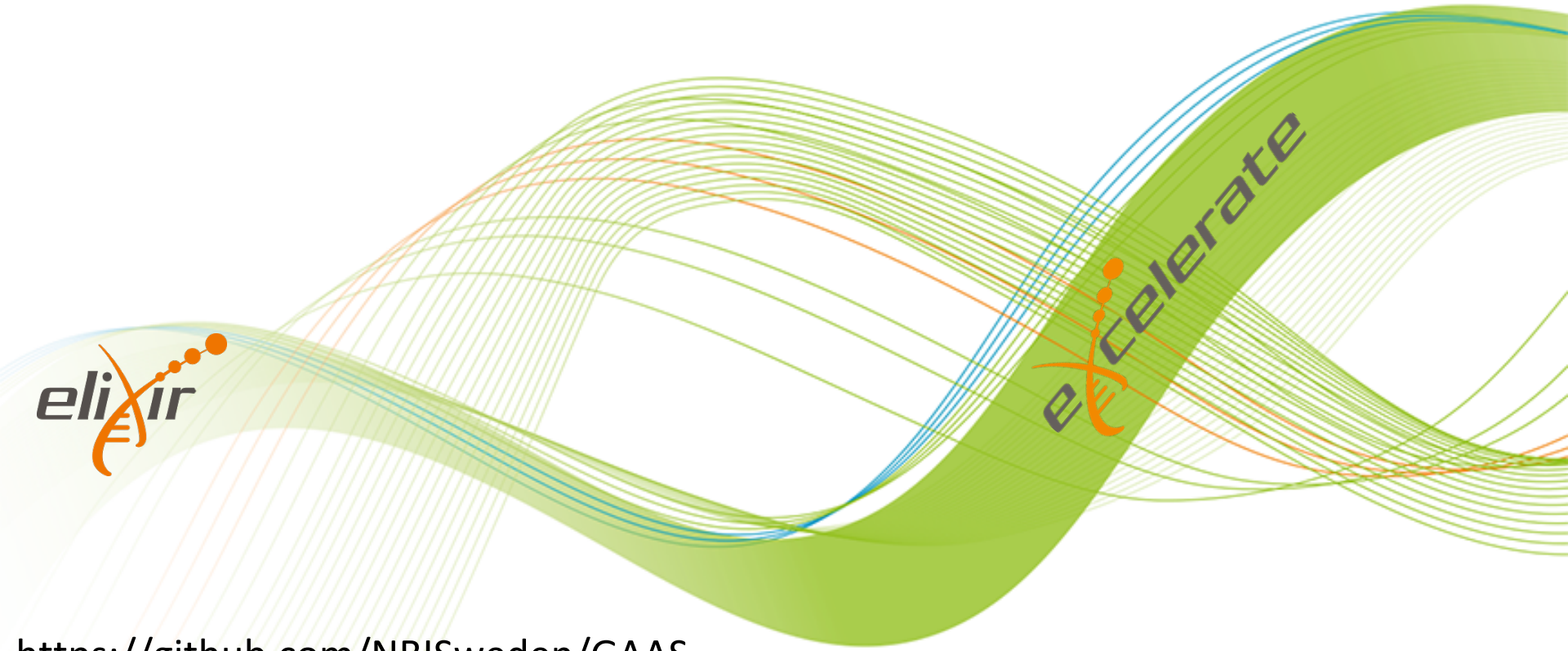
THE END



https://github.com/NBISweden/GAAS