

BRAKER1: Unsupervised RNA-Seq-Based Genome Annotation with GeneMark-ET and AUGUSTUS

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ABSTRACT

Motivation: Many genome sequencing projects are accompanied by transcriptome sequencing. GeneMark-ET is a gene prediction tool that incorporates unassembled RNA-Seq reads into unsupervised training and subsequently generates *ab initio* gene predictions. AUGUSTUS is a gene finder that usually requires supervised training and uses information from unassembled RNA-Seq reads in the prediction step.

Results: We present BRAKER1, a pipeline for unsupervised RNA-Seq-based genome annotation that combines the advantages of GeneMark-ET and AUGUSTUS. BRAKER1 requires an RNA-Seq read alignment file and a genome file as input. First, GeneMark-ET performs iterative training and generates initial gene structures. Second, AUGUSTUS uses predicted genes for training and then integrates RNA-Seq read information into final gene predictions. In our experiments, we observed that BRAKER1 was more accurate than MAKER2 when it is using RNA-Seq as sole source for training and prediction. BRAKER1 does not require pre-trained parameters or a separate training step.

Availability: BRAKER1 is available for download at <http://bioinf.uni-greifswald.de/downloads/> and <http://exon.gatech.edu/>.

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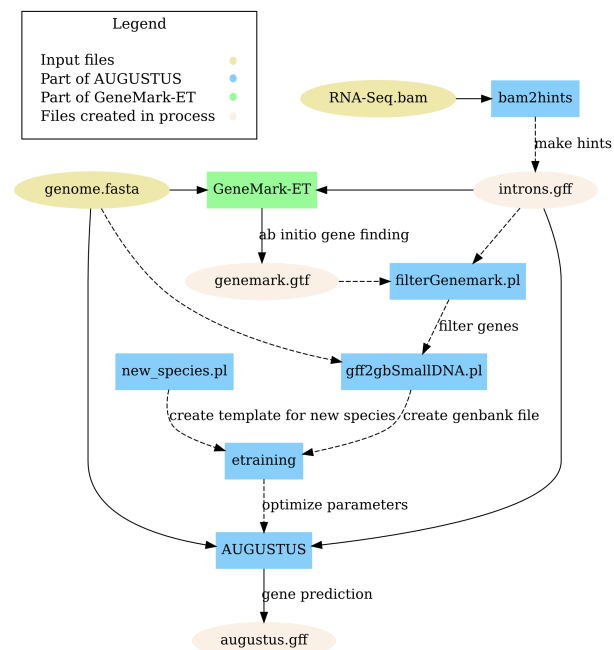


Fig. 1. Schematic view of the BRAKER1 pipeline.

1 INTRODUCTION

2 PIPELINE DESCRIPTION

BRAKER1 is implemented in Perl and requires two input files: an RNA-Seq alignment file in bam-format, and a corresponding genome file in fasta-format. Spliced alignment information is extracted from the RNA-Seq file and stored in gff-format. GeneMark-ET uses the genome file and the spliced alignment gff-file for RNA-Seq supported unsupervised

training. After training, GeneMark-ET creates an *ab initio* gene set. Those gene structures that have support by RNA-Seq alignments in all introns are selected for automated training of AUGUSTUS. After training, AUGUSTUS predicts genes in the input genome file using spliced alignment information from RNA-Seq as extrinsic evidence. The pipeline is illustrated in figure 1.

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3 TEST DATA

4 ACCURACY RESULTS

5 CONCLUSION

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