Abstract/Session Information for Program Number 1566W

Session Information

Session Title: Bioinformatics and Genomic Technology **Session Type:** Poster

Session Location: Exhibit Hall, Level 1, Convention Center **Session Time:** Wed 10:00AM-4:30PM

Abstract Information

Program Number: 1566W **Presentation Time:** Wed, Oct 23, 2013, 11:30AM-12:30PM

Keywords: Bioinformatics and Genomic Technology, KW151 - regulation of transcription, KW169 - transcription, KW155 - RNA, KW083 - genotype-phenotype correlations, KW008 - bioinformatics

Abstract Content

Detecting Differentially Expressed Genes in RNA-Seq Data with Unknown Conditions. *G. Klambauer, T. Unterthiner, S. Hochreiter* Institute of Bioinformatics, Johannes Kepler University Linz, Linz, Upper Austria, Austria.

Methods that identify differential expression in RNA-Seq data are currently limited to study designs in which two or more sample conditions are known a priori. However, these biological conditions like activated regulatory and metabolic pathways are typically unknown in genetic studies such as the HapMap or the 1000 Genomes project. We suggest DEXUS for detecting differential expression in RNA-Seq data for which the sample conditions are unknown. In a Bayesian framework DEXUS models read counts as a finite mixture of negative binomial distributions in which each mixture component corresponds to a condition. Evidence of differential expression is measured by the informative/non-informative (I/NI) value, which allows differentially expressed transcripts to be extracted at a desired specificity (significance level) or sensitivity (power). DEXUS performed excellently in identifying differentially expressed transcripts in data with unknown conditions. On 2,400 simulated data sets, I/NI value thresholds of 0.025, 0.05, and 0.1 yielded average specificities of 92%, 97%, and 99% at sensitivities of 76%, 61%, and 38% respectively. In cohorts with genetic and RNA-Seq data, DEXUS was able to detect differentially expressed transcripts that could be related to genetic variants via the identified conditions. These genetic variants can be classified into structural variants like copy number variations and single nucleotide variants, that is, eQTLs.

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