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View Reviews

Paper ID

1507

Paper Title

SPARSE NEGATIVE BINOMIAL SIGNAL RECOVERY FOR GENOMIC VARIANT PREDICTION IN DIPLOID SPECIES

Track Name

Main Track: Biomedical Imaging and Signal Processing

Reviewer #1

Questions

1. Importance/Relevance

3. Of sufficient interest

2. Novelty/Originality

3. Moderately original

3. Technical Correctness

3. Probably correct

4. Justification of Technical Correctness Score (required if score is 1 or 2)

I noticed following errors:

- Eq. 1: opening and closing parentheses and square brackets do not match.
- Eq. 8: optimization problem is defined over 6 variables all of which are scalar, but left hand side (f^{k+1}) is $6n$ dimensional vector. Authors should clarify this.
- Objective function in Eq. 8 is Q_j defined in previous equation. Using Q_j in Eq. 8 instead of rewriting the same thing will make it easier to follow the method.

5. Experimental Validation

1. Insufficient validation

6. Justification of Experimental Validation Score (required if score is 1 or 2)

Can authors clarify the following points for the result section?

- Why aren't the performance for heterozygous vectors given?
- Does the SPIRAL have hyperparameters, if so how are they selected?
- Does the SPIRAL also returns inherited and novel SVs for the child? I assume not as novel and inherited division is indicated to be novelty of the manuscript. If this is the case, how do they create center and right column of Fig.2 for SPIRAL.
- What value is used for p ?
- How do the authors choose λ_P and λ_C ?

7. Clarity of Presentation

3. Clear enough

8. Justification of Clarity of Presentation Score (required if score is 1 or 2)

It is hard to follow Section 2.3. Do authors describe a way to optimize Eq. 7? If so what does the last paragraph of Section 2.2 describe? Also, in step 1 of Section 2.3, there is a k . I assume this is not the same k as in Eq. 7. Can authors make Section 2.3 more clear.

9. Reference to Prior Work

3. References adequate

10. Overall evaluation of this paper

2. Marginal reject

11. Detailed assessment of the paper (seen by the authors):

The manuscript presents a SV prediction method from sequencing data. Compare to previous works, they try to handle noise in the data and consider novel and inherited SVs. For this, they model their data with negative binomial and predict SVs by likelihood maximization with an optimization procedure. The paper is mostly well written and my main concerns are about the results section as written above.

Reviewer #2

Questions

1. Importance/Relevance

2. Of limited interest

2. Justification of Importance/Relevance Score (required if score is 1 or 2)

The paper is focussed on a Bioinformatics problem/Genomic signal processing and would be of interest to a narrow section of the SP community.

3. Novelty/Originality

2. Minor originality

4. Justification of Novelty/Originality Score (required if score is 1 or 2)

The Poisson model in Willett's work (ref. [18] in the paper) is replaced with a negative binomial model -- this seems to be the main novelty. The penalty is also modified to include a scaling term for some

components using a weight factor $\gamma > 1$, in the penalty preceding Eq.(5).

The negative binomial distribution model must be justified thoroughly. Explanation must be provided to motivate as to why this would be a better model.

5. Technical Correctness

3. Probably correct

6. Justification of Technical Correctness Score (required if score is 1 or 2)

Order of square brackets and parentheses in equation (1) is flipped.

The parameters of the Negative Binomial distribution in equation (1) are arranged in two lines making it hard to read.

The equation ($f_{\{C\}} = z_{\{H\}} + y_{\{H\}} + z_{\{N\}} + y_{\{N\}}$) mentioned right before the second constraint in the subsection Familial Constraints seems to contradict the equation for the true signal ($f_{\{i\}} = 2z_{\{i\}} + y_{\{i\}}$) from the Methods section.

7. Experimental Validation

3. Limited but convincing

8. Justification of Experimental Validation Score (required if score is 1 or 2)

Validation reported on simulated data.

9. Clarity of Presentation

3. Clear enough

10. Reference to Prior Work

3. References adequate

11. Overall evaluation of this paper

4. Definite accept

12. Detailed assessment of the paper (seen by the authors):

The two main contributions of this paper are: The use of Negative Binomial model compared to earlier work which uses a Poisson-based model; and using a modified penalty which is not the vanilla l_1 penalty used in earlier work. Simulated data is used for evaluation. The proposed model performs better(ROC and Precision-Recall curves) than the SPIRAL model from Harmany et al.'s paper([18]).

Reviewer #3

Questions

1. Importance/Relevance

2. Of limited interest

2. Justification of Importance/Relevance Score (required if score is 1 or 2)

No related research has been conducted at this conference.

The reference papers mentioned in the introduction, excluding the papers written by the authors, are traditional algorithms.

3. Novelty/Originality

3. Moderately original

4. Technical Correctness

3. Probably correct

5. Experimental Validation

2. Lacking in some respect

6. Justification of Experimental Validation Score (required if score is 1 or 2)

Unlike the referenced papers [16] and [19], the experiment was conducted with limited data (simulation) and was not compared with

the latest algorithm.

7. Clarity of Presentation

2. Difficult to read

8. Justification of Clarity of Presentation Score (required if score is 1 or 2)

There are many notations that are not explained in the formula.

The last formula in chapter 2.1 is difficult to understand.

Several format (such as: Figure 1. -> Fig. 1.) do not conform to the ICASSP format.

9. Reference to Prior Work

3. References adequate

10. Justification of Reference to Prior Work Score (required if score is 1 or 2)

Several references do not conform to the IEEE format.

11. Overall evaluation of this paper

1. Definite reject

12. Justification of Overall evaluation of this paper (required if score is 1 or 2)

Although this study differs from previous studies written by the authors, the description of formulas makes it difficult for readers to accept the idea.

13. Detailed assessment of the paper (seen by the authors):

This is the first study to show that SVs can be predicted by analyzing related individuals, specifically a parent and child. They utilize a likelihood-based algorithms for predicting the most likely SVs present in each individual's genome. They further enforce sparsity in our predictions through an ℓ_1 penalty term.

Questions

1. Importance/Relevance

1. Irrelevant or out of scope

2. Novelty/Originality

2. Minor originality

3. Technical Correctness

2. Contains minor errors

4. Experimental Validation

2. Lacking in some respect

5. Clarity of Presentation

2. Difficult to read

6. Reference to Prior Work

2. References missing

7. Overall evaluation of this paper

2. Marginal reject

8. Detailed assessment of the paper (seen by the authors):

In this work, the authors proposed an optimization method for detecting both structural variants and their genotype (homozygous or heterozygous) from low-coverage DNA sequencing data in related individuals. The application is important and the results are promising. On the other hand, I struggle to see the scientific contribution of the work described in this document.