

CNV detection using RF Classifier

A PROJECT REPORT

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BONAFIDE CERTIFICATE

Certified that this project report “CNV DETECTION USING RF CLASSIFIER” is the bonafide work of PRASOON GOSWAMI, who carried out under my supervision.

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PROJECT GUIDE

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EXTERNAL EXAMINER

ABSTRACT

Copy number variants (CNV) are associated with phenotypic variation in several species. However, properly detecting changes in copy numbers of sequences remains a difficult problem, especially in lower quality or lower coverage next-generation sequencing data. Here, inspired by recent applications of machine learning in genomics, we describe a method to detect duplications and deletions in short-read sequencing data. In low coverage data, machine learning appears to be more powerful in the detection of CNVs than the gold-standard methods of coverage estimation alone, and of equal power in high coverage data. We also demonstrate how replicating training sets allows more precise detection of CNVs, even identifying novel CNVs in two genomes previously surveyed thoroughly for CNVs using long-read data.

List of tables, graphs and figures

FIGURES

- | | | |
|-----------------|-----------|-------------|
| ● Random forest | Chapter 3 | Section 3.1 |
| ● Scatter plots | Chapter 5 | Section 5.1 |
| ● Box plots | Chapter 5 | Section 5.2 |
| ● Heat map | Chapter 5 | Section 5.3 |

TABLES

- | | | |
|------------------------|-----------|-------------|
| ● Confusion matrix | Chapter 5 | Section 5.4 |
| ● RF classifier output | Chapter 5 | Section 5.5 |

Table of Contents

CHAPTER NO	TITLE	PAGE NO.
	Abstract	iv
	List of tables and figures	v
1.	Project Introduction	1
	1.1 Project objective	2
	1.2 Project description	2
	1.3 Hardware and software requirements	3
	1.4 Copy Number Variation	3
2.	Literature Review	5
3.	Introduction to RF Classifier	8
	3.1 Introduction	9
	3.2 Algorithm	9
	3.3 Advantages	10
	3.4 Disadvantages	10
4.	Design and Implementation	11
	4.1 The dataset	12
	4.2 Generating the dataset	12
	4.3 Method	14
	4.4 Algorithm implementation	15
5.	Results and Analysis	16
	5.1 Scatter Plot	17
	5.2 Box Plot	18
	5.3 Heat Map	19
	5.4 Confusion Matrix	20
	5.5 Classifier Output	22

Conclusion	23
Future Scope and Enhancement	24
References	25