#### FACULDADE DE ENGENHARIA DA UNIVERSIDADE DO PORTO

# Information and Data Analysis System for Gene Expression

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TECHNICAL REPORT



Mestrado Integrado em Engenharia Informática e Computação

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## **Abstract**

## Resumo

## Acknowledgements

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## **Contents**

1	Intr	oduction	1
	1.1	Context and Motivation	1
	1.2	Objectives	1
	1.3	Document Outline	1
2	Bacl	kground	3
	2.1	Introduction	3
	2.2	Genome Assembly and RNA Sequencing	3
		2.2.1 Assembly Methods	3
		2.2.2 RNA Sequencing Tools	3
		2.2.3 Common Data Formats	3
	2.3	Machine Learning	3
3	Data	asets and Validation	5
	3.1	Datasets	5
	3.2	Result Validation	5
4	Wor	rk Plan	7
	4.1	Development Phases	7
	4.2	Schedule Planning	7
Re	eferen	nces	9

#### CONTENTS

## **List of Figures**

#### LIST OF FIGURES

## **List of Tables**

#### LIST OF TABLES

### **Abbreviations**

cDNA Complementary DNA DNA Deoxyribonucleic Acid

FEUP Faculty of Engineering of the University of Porto(Faculdade de Engenharia

da Universidade do Porto)

IBMC Institute for Molecular and Cell Biology (Instituto de Biologia Molecular e

Celular)

mRNA Messenger RNA RNA Ribonucleic Acid RNA-Seq RNA Sequencing tRNA Transfer RNA

WTSS Whole Transcriptome Shotgun Sequencing

#### **Chapter 1**

#### Introduction

#### 1.1 Context and Motivation

#### 1.2 Objectives

#### 1.3 Document Outline

Besides the introduction chapter, this document is composed three additional chapters. These chapters have the following structure:

**Chapter 2** introduces some basic Biology and RNA Sequencing concepts, that are essential to understand the problems with which this document deals. Furthermore, we describe the main techniques used for genome/transcriptome sequencing and assembly, their differences and applications and the tools and data formats typically used on those areas. Lastly, we give some insight about machine learning algorithms and how they will be applied to this work.

**Chapter 3** presents the datasets that will be studied and used in this work, their origins and features. We will also refer the validation methods that will be used to access the quality of our results.

**Chapter 4** outlines the main steps in the development of this thesis (and the respective software prototype). In the last part of this chapter we will attempt to provide a feasible schedule for this work's execution.

#### Introduction

### Chapter 2

## Background

- 2.1 Introduction
- 2.2 Genome Assembly and RNA Sequencing
- 2.2.1 Assembly Methods
- 2.2.2 RNA Sequencing Tools
- 2.2.3 Common Data Formats
- 2.3 Machine Learning

#### Background

## Chapter 3

## **Datasets and Validation**

- 3.1 Datasets
- 3.2 Result Validation

#### Datasets and Validation

## Chapter 4

## **Work Plan**

- 4.1 Development Phases
- 4.2 Schedule Planning

#### Work Plan

## References