

# **Bioinformatics for Disease**

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# Preface

This is a Quarto book.

To learn more about Quarto books visit <https://quarto.org/docs/books>.

1 + 1

[1] 2

# **1 Molecular Sequencing Concepts and Technologies**

# **Part I**

## **Data skills**

In summary, this book has no content whatsoever.

**1 + 1**

[1] 2

## **2 Common data visualization techniques**



## **Part II**

# **DNA**

### **3 Aligning DNA sequences**

## 4 DNA quality control

Here I'm listing some useful reference material.

Chapter [14](#)

## **5 Variant calling**

## **Part III**

# **RNA**

## 6 Aligning RNA sequences

## **7 RNA quality control**

## **8 Differential expression analysis**



## **9 Gene set enrichment analysis (GSEA)**

**Part IV**

**Cancer Analyses**

## **10 Cancer drivers**

## **11 Driver enrichment**

## **12 Driver enrichment**

**Part V**

**Appendices**

Here I'm listing some useful reference material.

Chapter [14](#)

# 13 Reference versions

## 13.1 Downloading genome reference files

Species	Version	Command
human	GRCh38/hg38	wget https://hgdownload.soe.ucsc.edu/goldenPath/hg38/bigZips/hg38.fa.gz
human	GRCh37/hg19	wget https://hgdownload.soe.ucsc.edu/goldenPath/hg19/bigZips/hg19.fa.gz
mouse	GRCm39/mm39	wget https://hgdownload.soe.ucsc.edu/goldenPath/mm39/bigZips/mm39.fa.gz
mouse	GRCm38/mm10	wget https://hgdownload.soe.ucsc.edu/goldenPath/mm10/bigZips/mm10.fa.gz

## 13.2 Additional Resources

1. [Broad Institute](#)



# 14 Useful databases

## 14.1 Downloading genome reference files

Species	Version	Command
human	GRCh38/hg38	wget <a href="https://hgdownload.soe.ucsc.edu/goldenP">https://hgdownload.soe.ucsc.edu/goldenP</a>
human	GRCh37/hg19	wget <a href="https://hgdownload.soe.ucsc.edu/goldenP">https://hgdownload.soe.ucsc.edu/goldenP</a>
mouse	GRCm39/mm39	wget <a href="https://hgdownload.soe.ucsc.edu/goldenP">https://hgdownload.soe.ucsc.edu/goldenP</a>
mouse	GRCm38/mm10	wget <a href="https://hgdownload.soe.ucsc.edu/goldenP">https://hgdownload.soe.ucsc.edu/goldenP</a>

## 14.2 Additional Resources

1. [Broad Institute](#)

## References