

# Introduction to single-cell RNA-seq analysis - Data sets and analyses

November 2025

# Data sets

- ▶ Childhood acute lymphoblastic leukemia (cALL)
  - ▶ Caron et al. 2020
  - ▶ The most common pediatric cancer
  - ▶ Characterized by bone marrow lymphoid precursors that acquire genetic alterations, resulting in disrupted maturation and uncontrollable proliferation
  - ▶ Up to 85–90% of patients are cured
  - ▶ Others do not respond to treatment or relapse and die
  - ▶ **Aim:** characterise the heterogeneity of gene expression at the cell level, within and between patients
  - ▶ **cells:** Bone Marrow Mononuclear cells (BMMCs)

# Samples

Five types of sample are considered:

- ▶ B-ALL patients:
  - ▶ **'ETV6-RUNX1'**, or 't(12;21)', four patients
  - ▶ **'HHD'**, or 'High hyper diploid', two patients
- ▶ T-ALL patients
  - ▶ **'PRE-T'**, two patients
- ▶ Healthy controls,
  - ▶ **Healthy pediatric controls**, three individuals

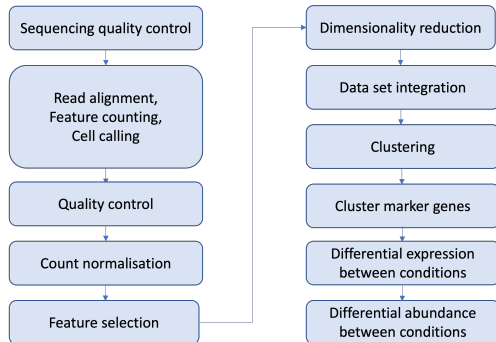
As the study aims at identifying cell populations, large numbers of cells were sequenced with the droplet-based 10x Chromium assay.

# Analyses

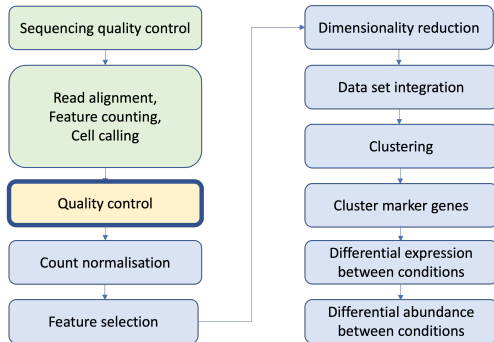
We will follow several steps:

- ▶ Sequencing quality check
- ▶ Alignment of reads to the human genome with 10x software cellranger
- ▶ Quality control (filter poor quality cells and remove uninformative genes)
- ▶ UMI count normalisation
- ▶ Feature selection and dimensionality reduction
- ▶ Data set integration
- ▶ Clustering
- ▶ Identification of cluster marker genes
- ▶ Differential expression and abundance between conditions

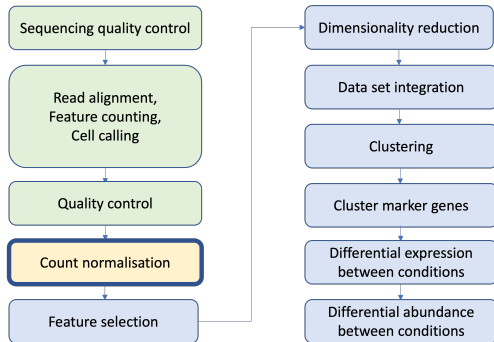
# Workflow



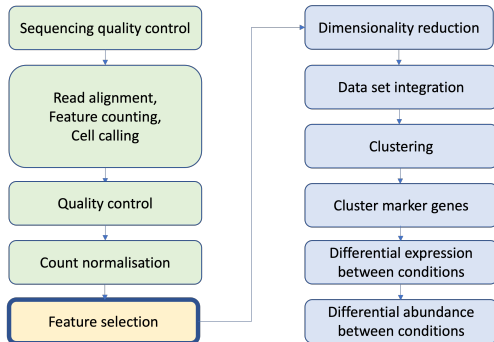
# Workflow



# Workflow

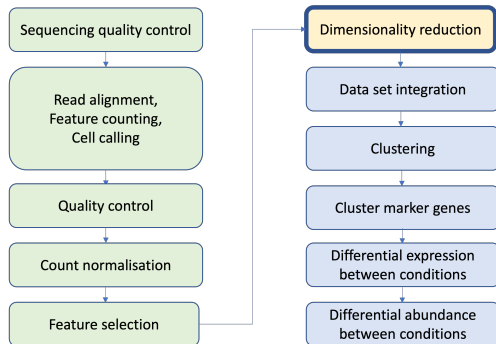


# Workflow

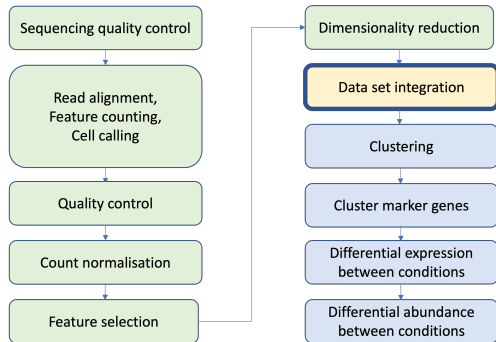




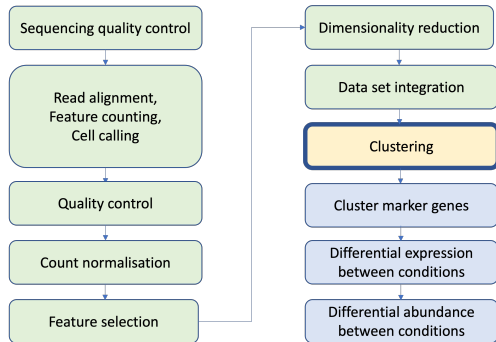
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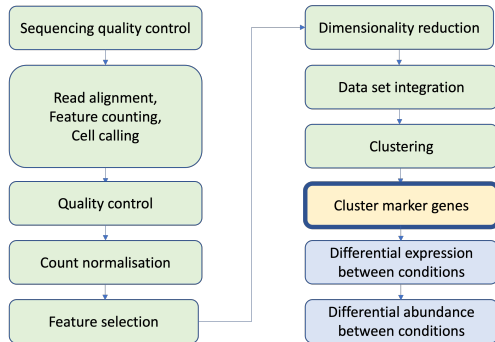
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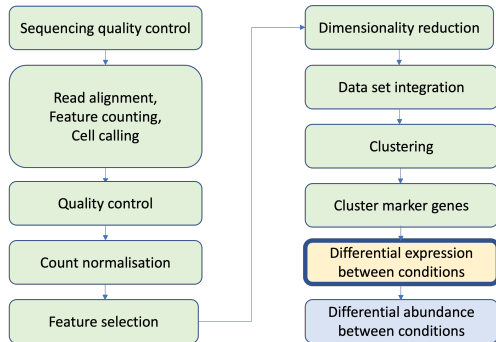
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