

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

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# Data set

## 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

# Samples

Four type of samples are considered:

- ▶ eight patients:
  - ▶ six B-ALL
    - ▶ four 't(12;21)' or 'ETV6-RUNX1'
    - ▶ two 'High hyper diploid' or 'HHD'
  - ▶ two T-ALL ('PRE-T')
- ▶ three healthy pediatric controls
- ▶ eight healthy adult controls, publicly available

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 (GRCh38) with 10X software cellranger
- ▶ quality control (cell calls, cells and genes filtering)
- ▶ UMI count normalisation
- ▶ data set integration (PBMMC and ETV6-RUNX1)
- ▶ feature selection and dimensionality reduction
- ▶ clustering
- ▶ marker gene identification
- ▶ differential expression and abundance between conditions
- ▶ trajectory analysis